



INTERNATIONAL APPLICATION PUBLISHED UNDER THE PATENT COOPERATION TREATY (PCT)

(51) International Patent Classification ⁶ : C12Q 1/68, C07K 16/18		A2	(11) International Publication Number: WO 99/64627
			(43) International Publication Date: 16 December 1999 (16.12.99)
(21) International Application Number: PCT/GB99/01780		(72) Inventor; and	
(22) International Filing Date: 4 June 1999 (04.06.99)		(75) Inventor/Applicant (for US only): ROBERTS, Gareth, Wyn [GB/GB]; The Grange, Church Street, Great Shelford, Cambs. CB2 5EL (GB).	
(30) Priority Data:		(74) Agent: DAVIES, Jonathan, Mark; Reddie & Grose, 16 Theobalds Road, London WCLX 8PL (GB).	
9812099.1	6 June 1998 (06.06.98)	GB	(81) Designated States: AE, AL, AM, AT, AU, AZ, BA, BB, BG, BR, BY, CA, CH, CN, CU, CZ, DE, DK, EE, ES, FI, GB, GD, GE, GH, GM, HR, HU, ID, IL, IN, IS, JP, KE, KG, KP, KR, KZ, LC, LK, LR, LS, LT, LU, LV, MD, MG, MK, MN, MW, MX, NO, NZ, PL, PT, RO, RU, SD, SE, SG, SI, SK, SL, TJ, TM, TR, TT, UA, UG, US, UZ, VN, YU, ZA, ZW, ARIPO patent (GH, GM, KE, LS, MW, SD, SL, SZ, UG, ZW), Eurasian patent (AM, AZ, BY, KG, KZ, MD, RU, TJ, TM), European patent (AT, BE, CH, CY, DE, DK, ES, FI, FR, GB, GR, IE, IT, LU, MC, NL, PT, SE), OAPI patent (BF, BJ, CF, CG, CI, CM, GA, GN, GW, ML, MR, NE, SN, TD, TG).
9813291.3	20 June 1998 (20.06.98)	GB	
9813611.2	24 June 1998 (24.06.98)	GB	
9813835.7	27 June 1998 (27.06.98)	GB	
9814110.4	1 July 1998 (01.07.98)	GB	
9814580.8	7 July 1998 (07.07.98)	GB	
9815438.8	16 July 1998 (16.07.98)	GB	
9815576.5	18 July 1998 (18.07.98)	GB	
9815574.0	18 July 1998 (18.07.98)	GB	
9816085.6	24 July 1998 (24.07.98)	GB	
9816086.4	24 July 1998 (24.07.98)	GB	
9816921.2	5 August 1998 (05.08.98)	GB	
9817097.0	7 August 1998 (07.08.98)	GB	
9817200.0	8 August 1998 (08.08.98)	GB	
9817632.4	14 August 1998 (14.08.98)	GB	
9817943.5	19 August 1998 (19.08.98)	GB	
(71) Applicant (for all designated States except US): GENOSTIC PHARMA LIMITED [GB/GB]; Sycamore Studios, New road, Over, Cambridge CB4 5PJ (GB).		Published With declaration under Article 17(2)(a). Without abstract; title not checked by the International Searching Authority.	
(54) Title: PROBES USED FOR GENETIC PROFILING			
<p style="text-align: right;">Pages:</p> <p style="text-align: right;">1-11</p> <p style="text-align: right;">35-44</p> <p style="text-align: right;">275-317</p> <p style="text-align: right;">394-416</p> <p style="text-align: right;">440-443</p>			

FOR THE PURPOSES OF INFORMATION ONLY

Codes used to identify States party to the PCT on the front pages of pamphlets publishing international applications under the PCT.

AL	Albania	ES	Spain	LS	Lesotho	SI	Slovenia
AM	Armenia	FI	Finland	LT	Lithuania	SK	Slovakia
AT	Austria	FR	France	LU	Luxembourg	SN	Senegal
AU	Australia	GA	Gabon	LV	Latvia	SZ	Swaziland
AZ	Azerbaijan	GB	United Kingdom	MC	Monaco	TD	Chad
BA	Bosnia and Herzegovina	GE	Georgia	MD	Republic of Moldova	TG	Togo
BB	Barbados	GH	Ghana	MG	Madagascar	TJ	Tajikistan
BE	Belgium	GN	Guinea	MK	The former Yugoslav Republic of Macedonia	TM	Turkmenistan
BF	Burkina Faso	GR	Greece			TR	Turkey
BG	Bulgaria	HU	Hungary	ML	Mali	TT	Trinidad and Tobago
BJ	Benin	IE	Ireland	MN	Mongolia	UA	Ukraine
BR	Brazil	IL	Israel	MR	Mauritania	UG	Uganda
BY	Belarus	IS	Iceland	MW	Malawi	US	United States of America
CA	Canada	IT	Italy	MX	Mexico	UZ	Uzbekistan
CF	Central African Republic	JP	Japan	NE	Niger	VN	Viet Nam
CG	Congo	KE	Kenya	NL	Netherlands	YU	Yugoslavia
CH	Switzerland	KG	Kyrgyzstan	NO	Norway	ZW	Zimbabwe
CI	Côte d'Ivoire	KP	Democratic People's Republic of Korea	NZ	New Zealand		
CM	Cameroon			PL	Poland		
CN	China	KR	Republic of Korea	PT	Portugal		
CU	Cuba	KZ	Kazakhstan	RO	Romania		
CZ	Czech Republic	LC	Saint Lucia	RU	Russian Federation		
DE	Germany	LI	Liechtenstein	SD	Sudan		
DK	Denmark	LK	Sri Lanka	SE	Sweden		
EE	Estonia	LR	Liberia	SG	Singapore		

PATENT COOPERATION TREATY

PCT

DECLARATION OF NON-ESTABLISHMENT OF INTERNATIONAL SEARCH REPORT


(PCT Article 17(2)(a), Rules 13ter.1(c) and Rule 39)

Applicant's or agent's file reference 39794/JMD	IMPORTANT DECLARATION	Date of mailing (day/month/year) 01/02/2000
International application No. PCT/GB 99/01780	International filing date (day/month/year) 04/06/1999	(Earliest) Priority date (day/month/year) 06/06/1998
International Patent Classification (IPC) or both national classification and IPC		C12Q1/68 C07K18/18
Applicant GENOSTIC PHARMA LIMITED.et.al		

This International Searching Authority hereby declares, according to Article 17(2)(a), that no international search report will be established on the international application for the reasons indicated below

1. ☐ The subject matter of the international application relates to:
- a. ☐ scientific theories.
 - b. ☐ mathematical theories
 - c. ☐ plant varieties.
 - d. ☐ animal varieties.
 - e. ☐ essentially biological processes for the production of plants and animals, other than microbiological processes and the products of such processes.
 - f. ☐ schemes, rules or methods of doing business.
 - g. ☐ schemes, rules or methods of performing purely mental acts.
 - h. ☐ schemes, rules or methods of playing games.
 - i. ☐ methods for treatment of the human body by surgery or therapy.
 - j. ☐ methods for treatment of the animal body by surgery or therapy.
 - k. ☐ diagnostic methods practised on the human or animal body.
 - l. ☐ mere presentations of information.
 - m. ☐ computer programs for which this International Searching Authority is not equipped to search prior art.
2. ☒ The failure of the following parts of the international application to comply with prescribed requirements prevents a meaningful search from being carried out:
- ☐ the description ☒ the claims ☐ the drawings
3. ☒ The failure of the nucleotide and/or amino acid sequence listing to comply with the standard provided for in Annex C of the Administrative Instructions prevents a meaningful search from being carried out:
- ☒ the written form has not been furnished or does not comply with the standard.
- ☒ the computer readable form has not been furnished or does not comply with the standard.
4. Further comments: see FURTHER INFORMATION sheet PCT/ISA/203

Name and mailing address of the International Searching Authority

 European Patent Office, P.B. 5818 Patentlaan 2
NL-2280 HV Rijswijk
Tel. (+31-70) 340-2040, Tx. 31 651 epo nl,
Fax: (+31-70) 340-3016

Authorized officer

Barbara Klaver



FURTHER INFORMATION CONTINUED FROM PCT/ISA/ 203

In view of the large number of alternative solutions to the obvious desirable objective of the provision of disease related nucleic acid and antibody probes, and in view of the wording of the claims as filed, it is impossible to determine the matter for which protection is sought. Accordingly, the present application fails to comply with the requirement of Article 6 PCT, first sentence (see also Rule 6.1(a) PCT) and fails to comply with the requirements for clarity and conciseness of Article 6 PCT, second sentence.

Moreover, the present claims relate to an extremely large number of possible undefined probes for which no technical features are provided. The claims cover all probes having the characteristic of being disease related, whereas the application provides no support at all within the meaning of Article 6 PCT and/or disclosure within the meaning of Article 5 PCT for such probes. In the present case, the claims so lack support, and the application so lacks disclosure, that a meaningful search is impossible. Independent of the above reasoning, the claims also lack clarity (Article 6 PCT). An attempt is made to define the probes by reference to a result to be achieved. Again, this lack of clarity in the present case is such so as to render a meaningful search impossible.

The sequence listing as present in the description does not comply with WIPO Standard ST 25 prescribed in the administrative instructions under Rule 5.2. Thus, the sequences have not been provided either on paper or machine readable form in accordance with the said instructions, and the Applicant has not remedied the disclosed deficiencies within the time limit fixed in the invitation to Rule 13ter.1a.

The applicant's attention is drawn to the fact that claims relating to inventions in respect of which no international search report has been established need not be the subject of an international preliminary examination (Rule 66.1(e) PCT). The applicant is advised that the EPO policy when acting as an International Preliminary Examining Authority is normally not to carry out a preliminary examination on matter which has not been searched. This is the case irrespective of whether or not the claims are amended following receipt of the search report or during any Chapter II procedure. If the application proceeds into the regional phase before the EPO, the applicant is reminded that a search may be carried out during examination before the EPO (see EPO Guideline C-VI, 8.5), should the problems which led to the Article 17(2) declaration be overcome.

predisposition to disease or a susceptibility to adverse drug responses (e.g. presenilin gene mutations and development of Alzheimer's disease, BRCA gene mutation and development of breast cancer, ACE polymorphisms and early onset heart disease, cytochrome P450 polymorphisms and drug metabolism).

However, such studies have been completed as academic exercises in scientific discovery and involve individual genes and large groups of patients.

Usually a particular individual response to disease or therapy is likely to result from a complex interaction between multiple genes, discrete environmental factors and the particular therapeutic approach offered (for example see algorithms in Figs. 1 and 2).

As a result, despite the many publications concerning the theoretical or potential applications of genomics to medicine (e.g. Marshall 1997a and b, Poste 1998, Crooke 1998), progress in implementing these approaches on a practical level has been exceedingly slow. In particular, little progress has been made in the understanding of or the ability to prognose individual response to particular disease states or therapeutic regimes (Poste 1998).

In part this has been related to the types of technology available for such studies (Marshall and Hodgson 1998). Such techniques as MALDI-TOF (Griffin et al 1997), sequencing (Dramanac et al 1998) and molecular beacons (Tyagi et al 1998) are complex and relatively slow and require the availability of specialised laboratories and highly trained personnel.

In recent reviews of the field it has been stated that:

- 'within next 10 years when not only all genes (will have been) identified but all common intragenic variation also' (Lander 1996).
- the 'assembly of comprehensive clinical databanks and their use for large-scale genetic association studies to define robust disease-gene risk correlations' constitutes a significant technological challenge (Poste 1998).
- 'if all human DNA variants were known this set would include all functional polymorphisms and if they could be analysed in all individuals comparison of phenotypes and correlation with genotype might make possible the assignment of function to every gene that predisposes to disease of any kind, and also to non-clinical phenotypes including behavioural traits. **The sheer task of this is overwhelming and may never be practical**' (Shafer and Hawkins 1998).

On the basis of the current state of the art it seems clear that translating the colossal investment in the human genome project into a means of revolutionising healthcare management requires both substantial creativity in the harnessing of technologies and considerable technical invention before its promise of can be realised.

For the realisation of the promised revolution in medicine two key factors require consideration;

- The human genome is made up of some 100,000 separate genes.
- Not all genes are of equal biological importance as regards the physiological functioning of humans.

The first issue, that of reading and tracking the volume of information encapsulated in the human genome by the sequence of 100,000 genes and their mutations and polymorphic variations, is beginning to be addressed by emergent technologies such as DNACHIPS, MALDI-TOF MS (Marshall and Hodgson 1998 see Table 1) and PEDIAT-type technologies (Fox 1998).

Table 1. The main features of some hybridization array formats currently available (Marshall & Hodgson 1998)

Company	Arraying method	Hybridization step	Readout	Main focus
Affymetrix (Santa Clara, CA)	On-chip photolithographic synthesis of -20-25-mer oligos onto silicon wafers, which are diced in 1.24 cm ² or 5.25 cm ² chips	10,000-260,000 oligo features probed with labelled 30-40 nucleotide fragments of sample cDNA or antisense RNA	Fluorescence	Expression profiling, polymorphism analysis, and diagnosis
Brax (Cambridge, UK)	Short synthetic oligo, synthesized off chip	1,000 oligos on a "universal chip" probed with tagged nucleic acids	Mass spectrometry	Diagnostics, expression profiling, novel gene identification
Hyseq (Sunnyvale, CA)	500-2000 nt DNA samples printed onto 0.6 cm ² (HyGnostics) or ~18 cm ² (Gene Discovery) membranes Prefabricated 5-mer oligos printed as 1.15 cm ² arrays onto glass (HyChip)	64 sample cDNA spots probed with 8,000 7-mer oligos (HyGnostics) or ≤55,000 sample cDNA spots probed with 300 7-mer oligos (Gene Discovery) Universal 1024 oligo spots probed 10 kb sample cDNAs, labelled 5-mer oligos and ligase	Radioisotope Fluorescence	Expression profiling, novel gene identification, and large-scale sequencing (Gene Discovery array), polymorphism analysis and diagnostics (HyGnostics/HyChip arrays), and large-sample sequencing (HyChip array)
Incyte Pharmaceuticals (Palo Alto, CA)	Piezoelectric printing for spotting PCR fragments and on-chip synthesis of oligos	≤ (eventually 10,000) oligo/PCR fragment spots probed with labelled RNA	Fluorescence and Radioisotope	Expression profiling Polymorphism analysis, Diagnostics
Molecular Dynamics (Sunnyvale, CA)	500-5000 nt cDNAs printed by pen onto ~10 cm ² on glass slide	~10,000 cDNA spots probed with 200-400 nt labelled sample cDNAs	Fluorescence	Expression profiling and novel gene identification
Nanogen (San Diego, CA)	Prefabricated ~20 mer oligos, captured onto electroactive spots on silicon wafer, which are diced. Into ≤ 1 cm ² chips	25, 64, 100, 400 (and eventually 10,000) oligo spots polarized to enhance hybridization to 200-400 nt labelled sample cDNAs	Fluorescence	Diagnostics and short tandem repeat identification
Protogene Laboratories (Palo Alto, CA)	On-chip synthesis of 40-50-mer oligos onto 9 cm ² glass chip via printing to a surface-tension array	≤8,000 oligo spots probed with 200-400 nt labelled sample nucleic acids	Fluorescence	Expression profiling, and polymorphism analysis
Sequenom (Hamburg, Germany and San Diego, CA)	Off-set printing of array, around 20-25-mer	250 locations per SpectroChip interrogated by laser desorption and mass spectrometry	Mass spectrometry	Novel gene identification, candidate gene validation, diagnostics, and mapping
Synteni (Fremont, CA)	500-5000 nt cDNAs printed by tip onto ~4	≤10,000 cDNA spots probed with 200-400 nt labelled sample	Fluorescence	Expression profiling and novel gene identification

	cm ² glass chip	cDNAs		
The German Cancer Institute (Heidelberg, Germany)	Prototypic DNA macrochip with on-chip synthesis of probes using f-moc or t-boc chemistry	Around 1000 spots on a 8x12 cm chip	Fluorescence/mass spectrometry	Expression profiling and diagnostics

These new technologies mark a significant advance in the potential application of genomic information to the problems of biology and human health. The reason for this is their capability of determining or confirming a large volume of DNA sequence data very quickly at the individual level. In this way they open the door to the application of genomic information to the individual patient.

These technologies are also evolving quickly according to Moore's Law (which posits that computer chips' power doubles every 18 months). For instance, three years ago the genechips made by leading companies held some 20,000 DNA probes. Currently genechips with 65,000 probes are available, and a chip with 400,000 probes has recently been produced (Marshall and Hodgson 1998). Applications for such technologies have included sequencing, diagnostics (mutation detection in the BRCA1 gene for cancer), gene discovery, gene expression profiling and gene mapping (Marshall and Hodgson 1998).

However despite their value as research and diagnostic tools, the genechips in existence are utilized largely as research tools (Marshall and Hodgson 1998). They have not been used as a tool for the express purpose of improving healthcare management by enabling the process of clinical prognosis and facilitating the generation of health risk profiles.

The reason for this is the failure to conceive of or invent an appropriate design which identifies the critical core of genes which are the most important in terms of human function. The genetic variability in this group of genes is the most important contributor to the variation in clinical and physiological phenotypes. Not all genes are equally important in the normal physiological functioning of the human body nor in the induction, development or progression of diseases or physiological states. In a given disease, as few as 5-10 genes in different configurations may be of seminal importance in determining the vast bulk of inter-individual variability to disease and therapeutic approaches (Drews 1997, Goodman and Gillman 1996).

As such, a device capable of delivering information on 10,000 genes may leave its user in grave danger of information overload and render him/her unable to identify and abstract the critical information required to enhance patient management or healthcare.

As a result, the translation of such technologies in genechip devices from research tools into healthcare management tools is severely limited (Marshall and Hodgson 1998, Poste 1998, Schafer and Hawkins 1997).

In an effort to overcome this difficulty a consortium of academic and industrial groups

(SNP Consortium) has been formed to try and identify the important disease related variants of human genes. The technologies to be used are the generation and assembly of a SNP map spanning the whole human genome and its application to linkage studies.

However, this approach is still in its infancy and is widely held to face considerable technical hurdles in the robust statistical analysis of huge datasets.

In order to bring about the integration of genomics into medical practice and enable design and building of a technology platform which will enable the everyday practice of molecular medicine a way must be invented for the DNA sequence data to be aligned with the identification of genes central to the induction, development, progression and outcome of disease or physiological states of interest:

Practitioners of molecular healthcare need to be able to;

- Identify the presence or absence of a selected group of genes and polymorphic variants central to the induction, development progression and outcome of disease or physiological states
- Focus on polymorphisms that lie within the coding or regulatory regions of the gene and are likely to result in altered structure or expression of the protein.
- Utilise the data on the core group of genes in order to generate guidelines and guidance for the healthcare management of patients or persons.

The invention described herein identifies the core group of genes required for the design development and manufacture of such a valuable aid to clinical management of the patient and general healthcare management.

According to the invention, the number of genes and their configurations (mutations and polymorphisms) needed to be identified in order to provide critical clinical information concerning individual prognosis is considerably less than the 100,000 thought to comprise the human genome.

The identification of the identity of the core group of genes enables the invention of a design for genetic profiling technologies which comprises of the identification of the core group of genes and their sequence variants required to provide a broad base of clinical prognostic information - 'genostics'.

By careful and lengthy research of the literature, tabulation of data, cross referencing of studies and conduction of a variety of experiments we have identified the core group of genes, which, if assessed for the presence of their functional variants, will enable an enhanced prognosis for an individual patient and form the basis for converting genetic profiling technologies from research tools into universal tools for health management.

Identification of the core group of genes and their functional variants also allows for said technologies to be utilised in generating individual health-risk profiles and profiling the health-risks of the population at large. The determination and

identification of sequence data required to identify the important functional variants is readily accomplished by those skilled in the practice of the relevant arts.

The invention does not provide a method for treatment as such. Nor does it provide a direct method of diagnosis of illness or health risk as such. Information obtainable using the invention can be used by a medical practitioner to tailor resources and therapy to meet the likely requirements of individual patients and selected populations of patients. For example in a complex regime or clinical management plan (as seen for example in Fig. 1 and 2) the invention allows the better prediction of the outcome of both the disease and the chosen therapeutic process.

The enablement of the invention and the generation of the information required for the design of 'genostics' requires:

1. Identification of sequence data (Example 1).
2. Assessment of the type and significance of sequence variation in the core group of genes (Examples 2,3,4).
3. Identification of likely genetic variation/disease relationships (Example 5 and 5a).
4. Means of identifying and detecting additional polymorphisms in the core group of genes (Example 6).
5. A practical approach to data analysis to generate information on prognosis (Example 7).
6. An illustration of how clinical management of a patient can be enhanced by utilising genetic profiling approaches (Example 8 and 9).

EXAMPLE 1

Gene sequence data is readily available in the public domain.

For the design of the GENOSTIC genechip device, gene sequence data can be retrieved, by persons skilled in the art, by searching the following public databases:

Website	Address	Description
DbEST	http://www.ncbi.nlm.nih.gov/dbEST	Database of expressed sequence tags
EBI/EMBL	http://www.ebi.ac.uk/mutations/	Mutations
EBI: The European Bioinformatics Institute, Hinxton, UK	http://www.ebi.ac.uk/ebi_home.html	Nucleotide Sequence Database
EMBL	http://www.ebi.ac.uk/queries/queries.html	Nucleotide Sequence Database
GDB: The Genome Database, Infobiogen European Node, FRANCE	http://www.gdb.org/gdb/gdbtop.html	Human Genome Database

GeneCards	http://bioinformatics.weizmann.ac.il/cards/index.html	GeneCards is a database of human genes, their products and their involvement in diseases.
GeneClinics	http://www.geneclinics.org/	<i>GeneClinics</i> (formerly <i>Genline</i>) is a knowledge base of expert-authored, up-to-date information relating genetic testing to the diagnosis, management, and counseling of individuals and families with inherited disorders.
Genethon	http://www.genethon.fr/genethon_en.html	The Human Genome Research Centre.
GSDB: Genome Sequence database	http://www.ncgr.org/	A collection of DNA sequence data and related information.
HGP: Human Genome Project Information	http://www.ornl.gov/TechResources/Human_Genome/home.html	Useful background & links.
Human Gene Mutation Database	http://www.uwcm.ac.uk/uwcm/mg/search	Mutations
NCBI	http://www.ncbi.nlm.nih.gov/	KEY SITE. Nucleotide Sequence retrieval start point.
OMIM: Online Mendelian Inheritance in Man	http://www.ncbi.nlm.nih.gov/Omim/	This database is a catalog of human genes and genetic disorders.
PubMed	http://www.ncbi.nlm.nih.gov/PubMed/	PubMed accesses MEDLINE medical literature database and links to full-text journals. It is also the literature component of the Entrez retrieval system for molecular biology information.
Research Tools (Science - NCBI)	http://www.ncbi.nlm.nih.gov/SCIENCE96/ResTools.html	A Gene Map of the Human Genome.
RHdb: Radiation Hybrid Database, Hinxton, UK	http://www.ebi.ac.uk/RHdb	Radiation Hybrid Database.
Stanford Human Genome Centre	http://www.shgc.stanford.edu/	Sequence database.
HUGO: The Human Genome Organisation	http://www.gene.ucl.ac.uk/hugo	HUGO is the international organisation of scientists involved in the Human Genome Project.
TIGR: The Institute for Genomic Research	http://www.tigr.org/	Genomic databases.
The National Human Genome Research Institute	http://www.nhgri.nih.gov/	Access to sequence databases

The Whitehead Institute Center for Genome Research	http://www.genome.wi.mit.edu/	Genome map and sequence information.
Unigene: Unique Human Gene Sequence Collection. (NCBI)	http://www.ncbi.nlm.nih.gov/UniGene/index.html	UniGene is a system for automatically partitioning GenBank sequences into a non-redundant set of gene- oriented clusters. Each UniGene cluster contains sequences that represent a unique gene, as well as related information such as the tissue types in which the gene has been expressed and map location.
University of Oklahoma	http://dna1.chem.ou.edu/index.html	Genomic databases
WEHI, Melbourne, Aus	http://wehih.wehi.edu.au/srs/srsc/	Sequence Retrieval System

Genes coding for proteins known to play a key role in organ function or disease are designated 'candidate genostic genes'. Variations within the gene structure may alter the regulatory or structural integrity of the gene product leading to enhancement or reduction in the specific function (e.g. receptor binding, enzyme activity). The exact role that a candidate gene plays in disease, prognosis and healthcare management can be fully ascertained by assessing the effects of variation in gene structure in particular patient groups, populations or individuals (see examples 2,3 and 4).

EXAMPLE 2 -Candidate Genostic Genes

Human Neuronal Nitric Oxide Synthetase

Gene Map Locus: 12q24.2q24.31(OMIM Ref. 163731).

One candidate 'genostic' gene is the gene encoding nitric oxide synthetase (NOS-1).

The enzymes responsible for NO synthesis in man constitute a family with at least three distinct isoforms: inducible, endothelial, and neuronal. Neuronal NO synthetase (NOS-1) is localised to human chromosome 12, and participates in diverse biologic processes including neurotransmission, the regulation of body fluid homeostasis, neuroendocrine physiology, control of smooth muscle motility, sexual function and monocyte biology.

Burnett et al. (1992) localized NO synthase to rat penile neurons innervating the corpora cavernosa and to neuronal plexuses in the adventitial layer of penile arteries. They demonstrated that small doses of NO synthase inhibitors abolished electrophysiologically induced penile erections establishing that nitric oxide is a physiologic mediator of erectile function.

Kharazia et al. (1994) found that all neurons in the striatum and many in the cortex were positive for nitric oxide synthase indicating a role of NOS in brain function.

NOS1 cDNA clones contain different 5-prime terminal exons spliced to a common exon 2. Xie et al. (1995) demonstrated that the unique exons are positioned within 300 bp of each other but separated from exon 2 by an intron that is at least 20 kb long. A CpG island engulfs the downstream 5-prime terminal exon. In contrast, most of the upstream exon resides outside of this CpG island. The upstream exon includes a GT dinucleotide repeat. The expression of these 2 exons is subject to transcriptional control by separate promoters. Nitric oxide is synthesized in skeletal muscle by neuronal-type NO synthase, which is localized to sarcolemma of fast-twitch fibers. Synthesis of NO in active muscle opposes contractile force. Brenman et al. (1995) showed that NOS1 partitions with skeletal muscle membranes owing to association of enzyme with dystrophin, the protein mutated in Duchenne muscular dystrophy. The dystrophin complex interacts with an N-terminal domain of NOS1 that contains a GLGF motif. Both humans with DMD and mdx mice show a selective loss of NOS1 protein and catalytic activity from muscle membranes. NOS1-deficient mice are resistant to neural stroke damage following middle cerebral artery ligation. Nelson et al. (1995) reported a large increase in aggressive behavior and excess, inappropriate sexual behavior in NOS1 'knockout' mice. Initial observations indicated that male (but not female) NOS1-deficient mice engaged in chronic aggressive behavior.

<i>Most</i>	Methyltransferase	✓
<i>Most</i>	Sulphotransferase	✓
<i>Most</i>	NADPH-cytochrome p450 reductase	✓

The inventory of drugs and preparations both registered and in development which can be matched to drug targets exhibiting genetic polymorphisms can be found in standard works of reference, in particular the British National Formulary, 1998, the Dental Practitioners' Formulary, 1998, Martindale, 1998, Herbal medicines, 1998. Drugs available in the United States can be found in U.S. Pharmacopeia, 1998, and drugs available in Japan can be found in Iryoyaku Nihon Iyakuhinshu, 1998, Ippanyaku Nihon Iyakuhinshu, 1998 and Hokenyaku Jiten, 1998. Drugs available in other countries can be found in the appropriate National Formularies. A list of drugs currently under development worldwide can be found in current journals and text (Pipeline pulse, 1999, Scrip, 1998, IDrugs, 1998, Current Opinion in Drug Discovery and Development, 1998).

The use of the Genostic approach described above would be of considerable utility in determining the likelihood and magnitude of therapeutic response to individual and combinations of drugs in the inventories described above. Such difficulties can arise from adverse events, variations in metabolism and drug-drug interactions in situations where several diseases, requiring treatment, exist in a given patient. The potential for adverse events or deleterious outcomes could be ascertained in individuals, patients or populations in relation to all of the drugs referred to above. These factors are of considerable importance in enabling the selection and monitoring of therapeutic interventions and effective healthcare management.

There are a number of different aspects to this invention. With the GENOSTIC™ approach, it would be possible to configure a different set of genes for each therapeutic area, across the whole of medicine, and for therapeutic intervention. The table below shows examples of the types of diseases included in each of the GENOSTIC™ therapeutic areas.

Therapeutic Area	Diseases
ADME	Drug absorption, distribution, metabolism & excretion (ADME), toxicity, responses to therapeutic intervention.
Oncology	Cancers, carcinomas, sarcomas, gliomas
Central Nervous System	Neurological (e.g. retinal disorders, multiple sclerosis), neuropsychiatric, psychiatric, psychological & social dysfunction, disease and damage.
Behavioural disturbance	Aggression, violent behaviour, anxiety, sleep disorder, attention deficit disorder, appetitive disorder, addiction, depression, bipolar affective disorder

Brain damage	Head injury, mental retardation, epilepsy, stroke, seizures, brain tumors
Dementia	Alzheimer's, Parkinson's, Huntington's, prion diseases, epilepsy, neurodegeneration,
Psychoses & personality	Schizophrenia, OCD, depression, bipolar affective disorder
Cardiovascular	Heart failure, hypertension, vasculitis, arrhythmia, cholesterolaemia, cardiomyopathy, atherosclerosis, valvular disease, coarctation, aneurysms, blood disorders, COPD.
Gastrointestinal	Gastric ulcers, duodenal ulcers, peptic ulcers, kidney disease, liver, pancreas, urinary, GERD (heartburn), nausea, diabetes mellitus, obesity
Respiratory	Lungs, anoxia, hypoxia, breathing problems, asthma, COPD, allergies
Immunity	Injury, inflammation, infection, AIDS
Development	Growth, differentiation, developmental disorders.
Skin, bone, muscle	Cornea disease, abnormal pigmentation, conductive hearing loss, arthritis, osteoporosis, myopathies, muscular atrophy, myositis, myoblastoma, eczema, dermatitis.
Metabolic & endocrine	Metabolism, reproduction, obesity Hormone action, diabetes
Headache	Migraine; trauma, infection
Sexual dysfunction	Infertility, impotency, male erectile dysfunction, female reproductive disorders

In a first aspect.

ADME (ABSORPTION, DISTRIBUTION, METABOLISM & ELIMINATION) & TOXICOLOGY

The invention relates to a method of assessing the most appropriate therapeutic intervention in an individual, patient, group or population suffering from the debilitating consequences of dysfunction, damage or disease of the body and its systems.

People vary enormously in their response to disease and also in their response to therapeutic interventions aimed at ameliorating the disease process and its progression. However, the provision of medical care and medical management is centered around observations and protocols developed in clinical trials on groups or cohorts of patients plan (Wetherall, Leadingham and Warrell 1996). This group data is used to derive a standardised method of treatment which is subsequently applied on an individual basis (e.g. the comment that drugs are often prescribed on the basis that everyone is an 70kg white male).

It is standard practice for clinicians to prescribe the same starting dose of a particular drug for a given indication and then adjust the treatment regimen by monitoring the progress of the disease and therapeutic response in individual patients. Observation of *actual* therapeutic outcome following these adjustments to patients therapy provides, the basis for determining a prognosis for the disease and developing a clinical management plan for patient care (eg. see Fig 1, algorithm for management of schizophrenia, from Fig 1 Taylor and Kerwin 1997, Fig 2 algorithm for treatment of depression from Fig 1 Pathare and Paton 1997).

The standard practice of clinical management has its disadvantages. In particular it is retro-active in that changes to patient management will occur following the emergence of therapeutic failures, adverse events or other difficulties in undertaking the therapeutic regime.

The toxicological effect of any treatment involves four main pathways, Absorption, Distribution, Metabolism and Elimination, better known as ADME. The most important axiom of toxicology is that "the dose makes the poison". Therefore variation in genes affecting the Absorption, Distribution, Metabolism and Elimination (ADME) of 'therapeutic' substances, accounts for much of the difference in individuals risk of toxicity.

Drugs interact with the body in many different ways to produce their effect. Some drugs act as false substrates of inhibitors for transport systems (e.g. calcium channels) or enzymes (acetylcholinesterase). Most drugs however, produce their effects by acting on receptors, usually located in the cell membrane, which normally respond to endogenous chemicals in the body (Weatherall, Leadingham and Warrell 1996). Drugs that activate receptors and produce a response are called agonists (e.g. cholinomimetics). Antagonists combine with receptors but do not activate them, thus reducing the probability of the transmitter substance combining with the receptor and

so blocking receptor activation. The ability of the drug to interact with the receptor depends on the specificity of the drug for the receptor or 'target' (Brody, Larner and Minneman 1998).

In addition to the main categories of agonist and antagonist drugs also have mechanisms of action which include:

- blockade of uptake or transport sites (e.g. selective serotonin reuptake inhibitors)
- enzyme inhibition (e.g. angiotensin converting enzyme inhibitors, acetylcholinesterase inhibitors)
- blockade of ion channels (calcium channel antagonists, anaesthetics)

Any drug may produce unwanted or unexpected adverse events, these can range from trivial (slight nausea) to fatal (aplastic anaemia). According to a recent article published in *JAMA* (Lazarou J, Pomeranz BH, Corey PN. 1998. Incidence of adverse drug reactions in hospitalised patients: a meta-analysis of prospective studies. *JAMA* Apr 15; 279 (15): 1200-5), in 1994, in US, 106,000 deaths were caused by adverse drug reactions, making ADRs the fourth leading cause of death in US. One of the main reasons for adverse events following drug intake is the drug binding to non-specific or non-target receptors in the body (Brody, Larner and Minneman 1998). Another reason is the interaction of the drug with other drugs given to the patient. This is a particular problem in the elderly who frequently suffer from multiple illnesses requiring many different classes of drugs and providing a real potential for drug interactions (Weatherall, Ledingham and Warrell 1996). The drug may also produce adverse events over time as the drug is absorbed, distributed, metabolised and excreted e.g. products of metabolising the drug may be reactive themselves and be toxic to the body. Being able to predicting the likelihood of particular individuals suffering from an adverse event and the severity of that event would be important tool for the practitioner.

Another problem the medical practitioner faces, is that certain patients may be particularly susceptible to drug addiction. Examples of drugs with known addictive properties are Amphetamines, Temazepam and Phenobarbitone, although having approved medicinal use e.g. phenobarbitone for epilepsy, they may cause problems of dependency and misuse in individuals. Knowledge of such an individual's susceptibility before prescribing certain drugs would be an advantage to the medical practitioner.

The core list of genes for the ADME Genostic, would prove of considerable value in aiding decisions concerning the appropriateness and relevance of therapeutic interventions using many drugs. The use of the ADME Genostic would be of considerable utility in determining the likelihood and magnitude of therapeutic response, complications from drug-drug interactions, the potential for adverse events and the difficulties that might arise due to previous, concurrent or future dysfunction, damage or disease of body systems in an individual, patient, group or population. All of these factors are of considerable importance in enabling the selection and monitoring of therapeutic interventions and effective healthcare management.

In addition, the core list of genes in the ADME genostic would also be of considerable

utility in enhancing the analysis of clinical trial data derived from drugs in development.

A list of drugs currently on the market can be found in standard works of reference, in particular the British National Formulary, 1998, the Dental Practitioners' Formulary, 1998, Martindale, 1998, Herbal medicines, 1998. Drugs available in the United States can be found in U.S. Pharmacopeia, 1998, and drugs available in Japan can be found in Iryoyaku Nihon Iyakuhinshu, 1998, Ippanyaku Nihon Iyakuhinshu, 1998 and Hokenyaku Jiten, 1998. Drugs available in other countries can be found in the appropriate National Formularies. A list of drugs currently under development worldwide can be found in current journals and text (Pipeline pulse, 1999, Scrip, 1998, IDrugs, 1998, Current Opinion in Drug Discovery and Development, 1998).

In a recent review entitled, 'Drug-metabolism research challenges in the new millenium: individual variability in drug therapy and drug safety', it has been stated that:

"with the rapid progress in the understanding of genetic polymorphism and the development of genechip technology, it becomes quite feasible for individuals to be genotyped with respect to critical genes targeted for drug intervention and genes essential for drug transport and metabolism.....the (future) objective is to identify key genetic variations that could impact drug response and drug safety." A.Y.H. Lu, (1998) *Drug metabolism and disposition*, Vol 26 (12) p1217-1222.

There is a wealth of information available on the genetic polymorphisms of enzymes involved in drug metabolism. Genetic variation in genes coding for proteins which act as drug metabolising enzymes, drug transporters, DNA repair enzymes, or drug targets can lead to the production of defective enzymes or altered receptor binding affinities. This can have profound effects on the drug efficacy, drug safety and optimal drug dosage. The genetic variation in these genes has been identified and is included in our ADME core list of genes.

The following tables give examples of genes in which polymorphisms are known to be associated with variation in response to drugs.

DRUG ABSORPTION

Drug	Drug-transporter, membrane protein	Polymorphic?
All	P-glycoprotein 1 (MDR1)	✓
All	P-glycoprotein 3 (MDR3)	✓

DRUG DISTRIBUTION

Drug	Drug-binding plasma protein	Polymorphic?
All	Serum albumin (ALB)	✓
All	Alpha 1 acid glycoprotein (AAG)	✓
All	Canalicular multispecific organic anion transporter (CMOAT or MRP2)	✓

All	Multidrug resistance associated protein (MRP1)	✓
All	Cytokine-suppressive antiinflammatory drug-binding protein 1 (CSBP1)	✓

DRUG METABOLISM

Drug	Drug-metabolising enzyme	Polymorphic?
All	Cytochrome P450 enzymes (CYP2C19; CYP2D6)	✓
All	UDP-glucuronosyltransferase	✓
All	N-acetyltransferase (NAT1)	✓
All	NADPH-cytochrome p450 reductase	✓

DRUG ELIMINATION

Drug	Drug-excretion protein	Polymorphic?
All	Bile salt export pump (BSEP)	✓
All	Sodium/bile acid cotransporter, (SLC10A1; SLC10A2)	✓

DRUG TARGETS FOR CNS MARKETING DRUGS

Drug	Drug Target	Polymorphic?
Tricyclic antidepressants (TCA)	Neurotransmitter (NA/5-HT) re-uptake proteins (NET & SERT)	✓
SSRIs	Selective serotonin transport re-uptake protein (SERT)	✓
MAOIs	monoamine oxidase A & B	✓
Benzodiazepines (GABA facilitators)/GABA antagonists. Barbiturates.	GABA receptors	✓
Beta-blockers	Noradrenaline (beta-adrenergic) receptors	✓
Atypical antidepressants	Alpha-adrenoceptors	✓
Beta-adrenoceptors antagonists	Beta-adrenoceptors	
Dopamine blockers/ boosters	Dopamine receptors	✓
Dopamine blockers/ boosters/depleters	Dopamine transporter (DAT1)	✓
Anticholinergics (muscarinic antagonists)	Muscarinic receptors	✓
Anticholinergics (nicotinic antagonists)	Nicotinic receptors	✓
Anticholinesterases	Acetylcholinesterase (ACHE)	✓
COMT inhibitor	Catechol-O-methyltransferase (COMT)	✓
Sodium channel blocker	Sodium channel	✓

Opioid analgesics & antagonists	Opioid receptors (OPRM1; OPRK1; OPRD1)	✓
Antipsychotics/neuroleptics (5-HT/D2 antagonists)	5-HT/D2 receptors	✓
Antiinflammatory drugs	Cyclooxygenase (COX1, COX2)	✓
Antihistamines	Histamine receptors	✓

DRUG TARGETS FOR CNS DRUGS IN DEVELOPMENT

Drug	Drug Target	Polymorphic?
Selective NAT inhibitors (SNRIs)	Noradrenaline transport reuptake protein (NAT1 or NET)	✓
5-HT1A-agonist	5-HT1A receptor (HTR1A)	✓
Selective 5-HT2A antagonist	5-HT2A receptor (HTR2A)	✓
Clozapine (MAOI)	5-HT2C receptor (HTR2C)	✓
Glycine antagonist	Glycine receptor (GLRA2)	✓
Cannabinoid receptor agonist (THC)	Cannabinoid receptor (CNR1)	✓
Calcium channel blocker	Calcium channels	✓

DRUG TARGETS FOR CARDIOVASCULAR MARKETED DRUGS

Drug	Drug Target	Polymorphic?
ACE inhibitors	Angiotensin converting enzyme (ACE)	✓
HMG CoA reductase inhibitors, e.g simvastatin	HMG CoA reductase	✓
Angiotensin II antagonists	Angiotensinogen (AGT)	✓
Calcium channel blocker	Calcium channel	✓
Thromboxane A2 synthase inhibitor	Thromboxane A2 synthase	✓
A2 receptor antagonist	Thromboxane A2 receptor	✓
Potassium channel blocker	Potassium channel	✓
Na-H ion exchange (NHE) inhibitor	Na-H ion exchanger (NHE)	✓
bile acid transport inhibitor	SLC10A1 (sodium/bile acid cotransporter)	✓
bile acid transport inhibitor	SLC10A2 (sodium/bile acid cotransporter)	✓
platelet aggregation inhibitor	Von Willebrand factor	✓
ACAT inhibitor	Acetoacetyl-CoA-thiolase (ACAT)	✓
Endothelin antagonist	Endothelin (EDN3)	✓

DRUG TARGETS FOR GASTROINTESTINAL DISEASE (Peptic ulcer) MARKETED DRUGS

Drug	Drug Target	Polymorphic?
------	-------------	--------------

Proton pump inhibitor (e.g. omeprazole).	H ⁺ /K ⁺ adenosine triphosphatase (ATPase) enzyme system ('proton pump')	✓
H ₂ antagonists (e.g. cimetidine)	Histamine H ₂ -receptor	✓
Muscarinic antagonists (e.g. pirenepine)	Muscarinic m1 & m3 receptors	✓
Prostaglandins (inhibit cAMP)	Adenylate cyclase, histamine-induced activity	✓

DRUG TARGETS FOR RESPIRATORY DISEASE (Asthma & Allergy) MARKETING DRUGS

Drug	Drug Target	Polymorphic?
Beta-2- agonists (Bronchodilators)	Beta-2-adrenoceptor	✓
Muscarinic antagonists (Bronchodilators)	Muscarinic receptors	✓
Histamine antagonists (Antihistamines)	Histamine receptors	✓
Thromboxane A ₂ synthase inhibitor	Thromboxane A ₂ synthase	✓
A ₂ receptor antagonist	Thromboxane A ₂ receptor	✓

DNA REPAIR

Drug	DNA repair enzyme	Polymorphic?
All	O(6)-methylguanine-DNA methyltransferase (MGMT)	✓
All	DNA damage binding protein (DDB1)	✓
All	DNA-damage-inducible transcript 3 (DDIT3)	✓
All	RAD52	✓

We have elaborated on the value and utility to be derived from the gathering together of the genes which form the core gene list for this particular Genostic system.

These genes are elaborated below:

KEY TO 'PROTEIN FUNCTION' COLUMN

- E ENZYME
- T TRANSPORT & STORAGE
- S STRUCTURAL
- I IMMUNITY
- N NERVOUS TRANSMISSION
- G GROWTH & DIFFERENTIATION

ADME GENE LIST

	HUGO gene symbol	Protein function
5-adenosyl homocysteine hydrolase		E
Acetoacetyl 1-CoA-thiolase	ACAT1	E
Acetoacetyl 2-CoA-thiolase	ACAT2	E
Acetyl CoA acyltransferase	ACAA	E
Acetylcholine receptor, nicotinic, alpha A1	CHRNA1	N
Acetylcholine receptor, nicotinic, alpha A2	CHRNA2	N
Acetylcholine receptor, nicotinic, alpha A3	CHRNA3	N
Acetylcholine receptor, nicotinic, alpha A4	CHRNA4	N
Acetylcholine receptor, nicotinic, alpha A5	CHRNA5	N
Acetylcholine receptor, nicotinic, alpha A6	CHRNA6	N
Acetylcholine receptor, nicotinic, alpha A7	CHRNA7	N
Acetylcholine receptor, nicotinic, beta 1	CHRNA1	N
Acetylcholine receptor, nicotinic, beta 2	CHRNA2	N
Acetylcholine receptor, nicotinic, beta 3	CHRNA3	N
Acetylcholine receptor, nicotinic, beta 4	CHRNA4	N
Acetylcholine receptor, nicotinic, epsilon	CHRNA5	N
Acetylcholine receptor, nicotinic, gamma	CHRNA6	N
Acetylcholinesterase	ACHE	E
Actin, alpha, cardiac	ACTC	S
Actin, alpha, skeletal	ACTA1	S
Actin, alpha, smooth, aortic	ACTA2	S
Actin, beta	ACTB	S
Actin, gamma 2	ACTG2	S
Acyl CoA dehydrogenase, short chain	ACADS	E
Adenine phosphoribosyltransferase	APRT	T
Adenosine deaminase	ADA	E
Adenosine monophosphate deaminase	AMPD	E
Adenosine receptor A1	ADORA1	N
Adenosine receptor A2A	ADORA2A	N
Adenosine receptor A2B	ADORA2B	N
Adenosine receptor A3	ADORA3	N
Adenylate cyclase 1	ADCY1	E
Adenylate cyclase 2	ADCY2	E
Adenylate cyclase 3	ADCY3	E
Adenylate cyclase 4	ADCY4	E
Adenylate cyclase 5	ADCY5	E
Adenylate cyclase 6	ADCY6	E
Adenylate cyclase 7	ADCY7	E
Adenylate cyclase 8	ADCY8	E
Adenylate cyclase 9	ADCY9	E
Adenylate kinase	AK1	E
Adenylate transferase		E
Adenylosuccinate lyase	ADSL	E
ADP-ribosyltransferase	ADPRT	E
Adrenergic receptor, alpha1	ADRA1	N
Adrenergic receptor, alpha2	ADRA2	N

Adrenergic receptor, beta1	ADRB1	N
Adrenergic receptor, beta2	ADRB2	N
Adrenergic receptor, beta3	ADRB3	N
Adrenocorticotrophic hormone (ACTH) receptor	ACTHR	G
Adrenoleukodystrophy gene	ALD	E
Albumin, ALB	ALB	T
Alkaptonuria gene	AKU	G
Alpha 1 acid glycoprotein	AAG; AGP	T
alpha1-antitrypsin	PI	E
alpha2-antiplasmin	PLI	E
alpha-amylase		E
Alpha-fetoprotein	AFP	G
alpha-glucosidase, neutral AB	GANAB	E
alpha-glucosidase, neutral C	GANC	E
Aminomethyltransferase	AMT	E
Aminopeptidase P	XPNPEP2	E
Amyloid beta (A4) precursor protein-binding, APBB1	APBB1	N
Amyloid beta A4 precursor protein	APP	N
Androgen binding protein	ABP	T
Androgen receptor	AR	G
Angiotensin converting enzyme	ACE, DCP1	E
Angiotensin receptor 1	AGTR1	T
Angiotensin receptor 2	AGTR2	T
Angiotensinogen	AGT	E
Annexin 1	ANX 1	I
Apurinic endonuclease	APE	E
Arginine vasopressin	AVP	N
Arginine vasopressin receptor 1A	AVPR1A	N
Arginine vasopressin receptor 1B	AVPR1B	N
Arginine vasopressin receptor 2	AVPR2	N
Aryl hydrocarbon receptor	AHR	T
Arylsulfatase E	ARSE	E
Aspartate transcarbamoylase		E
Ataxia telangiectasia gene, AT	ATM	G
ATP cobalamin adenosyltransferase		E
ATP sulphurylase	atpsk2	E
ATP/ADP translocase		E
Atrial natriuretic peptide	ANP	G
Atrial natriuretic peptide receptor A	NPR1	G
Atrial natriuretic peptide receptor B	NPR2	G
Atrial natriuretic peptide receptor C	NPR3	G
BCL2-associated X protein	BAX	G
Benzodiazepine receptor		N
beta-endorphin receptor		N
Bile acid coenzyme A: amino acid N-acyltransferase	BAAT	E

In a twelfth aspect.

DEVELOPMENT

The present invention relates to a method of assessing the risk of developing clinical or social consequences following dysfunction, damage or disease of the body consequent to an aberration in the processes of development and indicating appropriate therapeutic interventions.

The process by which fertilisation of an egg leads to the formation and growth of a foetus, birth of a baby and the maturation of an adolescent into an adult are collectively described as development. An understanding of the genetic and molecular events directing the development and differentiation of cells into tissues and organs is slowly being understood (Gilbert 1997). The intricate nature of the interactions between cells as they divide and differentiate is mediated by a host of regulatory systems including:

DNA methylation

Transcriptional regulation (e.g POU transcription factors)

Differential RNA splicing

Paracrine systems

Signal transduction pathways (e.g. RTK-Ras, JAK-STAT, NOTCH)

Neurotransmitter/receptor interaction

Cell surface adhesion molecules

In addition there are significant interactions between the developing organism and the environment (the womb and subsequently the external environment). In humans the process of development and maturation continues through to late 20's as the final stages of brain myelination occur.

The sheer complexity of these interactions and their subtle effects on the dynamics of organ formation and development mean that there are multiple opportunities for perturbation, failure or premature termination of the developmental trajectory. No tissue, organ or organ system in the body is immune to the possibility of dysfunction, damage or disease consequent to an aberration in the processes of development.

The spectrum of medical, psychological and social consequences consequent to an aberration in developmental processes is enormous (Weatherall, Leadingham and Warrell 1996). For example abnormalities of brain development are very frequent and often lead to lasting impairments in cognition and learning (some 3% of school leavers may have some degree of neurological impairment. Developmental disorders include:

Down's syndrome (brain and other organs)

Cruzon syndrome (skull)

Congenital adrenal hyperplasia (endocrine system)

Congenital hypothyroidism (endocrine system)

Hirschsprung's disease (gastrointestinal system)

Pyloric stenosis (gastrointestinal system)

Aortic-valve stenosis (cardiovascular system)

Mitral valve abnormalities (cardiovascular system)

Spina bifida (spine)

Cerebral palsy (central nervous system)

Cystic fibrosis (respiratory system)

The physiology and nature of dysfunction, damage or disease of the body consequent to an aberration in the processes of development are extremely complex. The exact spectrum of symptoms and attendant disability are derived from the nature of the lesion, its site and extent and the time at which it influenced the pattern of development. The presence of a clinical, psychological or social liability may also change over time since the manifestations of the difficulties at birth, adolescence or adulthood will alter as a function of the unfolding of development.

The interactions between the various proteins which form the constituent parts of the regulatory systems are critical in the control and modulation of development. Variation in the functionality of the proteins involved in these processes will, inevitably, cause or have an impact on the functioning of these systems or modulate a tissue's ability to minimise developmental aberrations and restore function following dysfunction, damage or disease in the development of these systems. A number of constitutional factors are known to impact on the individual's ability to deal with and recover from dysfunction, damage or disease of the body consequent to an aberration in the processes of development. These include genetic history, age, sex, nutritional status, pre-existing disease or injury, drug treatments and socio-economic circumstances.

Genetic variation within individuals is also a key factor although the extent and nature of the genes involved and their precise impact on prognosis, complications, efficacy of therapeutic intervention and eventual recovery of function is largely unknown.

The individual variability in response to the occurrence of dysfunction, damage or disease of the body consequent to an aberration in the processes of development and the associated variation in symptomatology, response to therapy and adverse events resulting from therapeutic interventions lies at the heart of the difficulties experienced in the health and social management of dysfunction, damage or disease of the body consequent to an aberration in the processes of development.

We have elaborated on the value and utility to be derived from the gathering together of the genes which form the core gene list for this particular Genostic system.

These genes are elaborated below:

KEY TO 'PROTEIN FUNCTION' COLUMN

E ENZYME
 T TRANSPORT & STORAGE
 S STRUCTURAL
 I IMMUNITY
 N NERVOUS TRANSMISSION
 G GROWTH & DIFFERENTIATION

DEVELOPMENT GENE LIST

	HUGO gene symbol	Protein function
17-ketosteroid reductase		N
2,4-dienoyl CoA reductase	DECR	E
3 beta hydroxysteroid dehydrogenase 2	HSD3B2	E
3-oxoacid CoA transferase	OXCT	E
6-pyruvoyltetrahydropterin synthase	PTS	E
Absent in melanoma 1 gene	AIM1	G
Acetoacetyl 2-CoA-thiolase	ACAT2	E
Acetyl CoA acyltransferase	ACAA	E
Acetyl CoA carboxylase alpha	ACACA	E
Acetylcholine receptor, nicotinic, alpha A1	CHRNA1	N
Acetylcholine receptor, nicotinic, alpha A2	CHRNA2	N
Acetylcholine receptor, nicotinic, alpha A3	CHRNA3	N
Acetylcholine receptor, nicotinic, alpha A4	CHRNA4	N
Acetylcholine receptor, nicotinic, alpha A5	CHRNA5	N
Acetylcholine receptor, nicotinic, alpha A6	CHRNA6	N
Acetylcholine receptor, nicotinic, alpha A7	CHRNA7	N
Acetylcholine receptor, nicotinic, beta 1	CHRNA1	N
Acetylcholine receptor, nicotinic, beta 2	CHRNA2	N
Acetylcholine receptor, nicotinic, beta 3	CHRNA3	N
Acetylcholine receptor, nicotinic, beta 4	CHRNA4	N
Acetylcholine receptor, nicotinic, epsilon	CHRNA5	N
Acetylcholine receptor, nicotinic, gamma	CHRNA6	N
Acetylcholinesterase	ACHE	E
Achromatopsia 2	ACHM2	S
Acid phosphatase 2, lysosomal	ACP2	E
Acrosin	ACR	G
Actin, alpha, cardiac	ACTC	S
Actin, alpha, skeletal	ACTA1	S
Actin, alpha, smooth, aortic	ACTA2	S
Activin		G
Activin A receptor, type 2B	ACVR2B	G
Activin A receptor, type 2-like kinase 1	ACVRL1	G
Acyl CoA dehydrogenase, short chain	ACADS	E
Acyl-CoA thioesterase		E
ADAM (A disintegrin and metalloproteinase) 1	ADAM1	E
ADAM (A disintegrin and metalloproteinase) 10	ADAM10	E
ADAM (A disintegrin and metalloproteinase) 11	ADAM11	E

ADAM (A disintegrin and metalloproteinase)	12	ADAM12	E
ADAM (A disintegrin and metalloproteinase)	13	ADAM13	E
ADAM (A disintegrin and metalloproteinase)	14	ADAM14	E
ADAM (A disintegrin and metalloproteinase)	15	ADAM15	E
ADAM (A disintegrin and metalloproteinase)	16	ADAM16	E
ADAM (A disintegrin and metalloproteinase)	17	ADAM17	E
ADAM (A disintegrin and metalloproteinase)	18	ADAM18	E
ADAM (A disintegrin and metalloproteinase)	19	ADAM19	E
ADAM (A disintegrin and metalloproteinase)	2	ADAM2	E
ADAM (A disintegrin and metalloproteinase)		ADAM3A	E
ADAM (A disintegrin and metalloproteinase)		ADAM3B	E
ADAM (A disintegrin and metalloproteinase)	4	ADAM4	E
ADAM (A disintegrin and metalloproteinase)	5	ADAM5	E
ADAM (A disintegrin and metalloproteinase)	6	ADAM6	E
ADAM (A disintegrin and metalloproteinase)	7	ADAM7	E
ADAM (A disintegrin and metalloproteinase)	8	ADAM8	E
ADAM (A disintegrin and metalloproteinase)	9	ADAM9	E
Adducin, alpha		ADD1	S
Adducin, beta		ADD2	S
Adenomatous polyposis coli tumour suppressor gene		APC	G
Adenosine deaminase		ADA	E
Adenosine monophosphate deaminase		AMPD	E
Adenosine receptor A1		ADORA1	N
Adenosine receptor A2A		ADORA2A	N
Adenosine receptor A2B		ADORA2B	N
Adenosine receptor A3		ADORA3	N
Adenyl cyclase			N
Adenylate cyclase 1		ADCY1	E
Adenylate cyclase 2		ADCY2	E
Adenylate cyclase 3		ADCY3	E
Adenylate cyclase 4		ADCY4	E
Adenylate cyclase 5		ADCY5	E
Adenylate cyclase 6		ADCY6	E
Adenylate cyclase 7		ADCY7	E
Adenylate cyclase 8		ADCY8	E
Adenylate cyclase 9		ADCY9	E
Adenylosuccinate lyase		ADSL	E
ADP-ribosyltransferase		ADPRT	E
Adrenergic receptor, alpha1		ADRA1	N
Adrenergic receptor, alpha2		ADRA2	N
Adrenergic receptor, beta1		ADRB1	N
Adrenergic receptor, beta2		ADRB2	N
Adrenergic receptor, beta3		ADRB3	N
Adrenocorticotrophic hormone (ACTH) receptor		ACTHR	G

Adrenoleukodystrophy gene	ALD	E
Alanine-glyoxylate aminotransferase	AGXT	E
Albumin, ALB	ALB	T
Aldehyde dehydrogenase 1	ALDH1	E
Aldehyde dehydrogenase 10	ALDH10	E
Aldehyde dehydrogenase 2	ALDH2	E
Aldehyde dehydrogenase 5	ALDH5	E
Aldehyde dehydrogenase 6	ALDH6	E
Aldehyde dehydrogenase 7	ALDH7	E
Aldolase A	ALDOA	E
Aldolase B	ALDOB	E
Aldolase C	ALDOC	E
Aldosterone receptor	MLR	E
Alkaline phosphatase, liver/bone/kidney	ALPL	G
Alkaptonuria gene	AKU	T
Alkylglycerone phosphate synthase	AGPS	G
Alpha 2 macroglobulin	A2M	E
alpha tectorin	TECTA	I
alpha thalassemia gene	ATRX	G
alpha1-antitrypsin	PI	N
alpha2-antiplasmin	PLI	E
alpha-actinin 2	ACTN2	E
alpha-actinin 3	ACTN3	G
alpha-amylase		G
Alpha-fetoprotein	AFP	E
alpha-Galactosidase A	GLA	G
alpha-ketoglutarate dehydrogenase		E
alpha-L-Iduronidase	IDUA	E
alpha-synuclein	SNCA	N
Amelogenin	AMELX	S
Aminomethyltransferase	AMT	E
Aminopeptidase P	XPNPEP2	E
Amphiregulin	AREG	G
Amylo-1,6-glucosidase	AGL	E
Amyloid beta (A4) precursor protein-binding, APBB1	APBB1	N
Amyloid beta A4 precursor protein	APP	N
Amyloid beta A4 precursor-like protein	APLP	N
Androgen binding protein	ABP	T
Androgen receptor	AR	G
Angiopoietin 1	ANGPT1	G
Angiopoietin 2	ANGPT2	G
Angiotensin converting enzyme	ACE, DCP1	E
Angiotensinogen	AGT	E
Ankyrin 1	ANK1	S
Ankyrin 2	ANK2	S
Ankyrin 3	ANK3	S
Antidiuretic hormone receptor	ADHR	T

Anti-Mullerian hormone	AMH	G
Anti-Mullerian hormone type 2 receptor	AMHR2	G
Antithrombin III	AT3	E
AP-2, alpha	TFAP2A	G
AP-2, beta	TFAP2B	G
AP-2, gamma	TFAP2C	G
Apaf-1		S
Apical protein, xenopus laevis-like	APXL	G
Apolipoprotein A 4	APOA4	T
Apolipoprotein A I	APOA1	T
Apolipoprotein A II	APOA2	T
Apolipoprotein B	APOB	T
Apolipoprotein C1	APOC1	T
Apolipoprotein C2	APOC2	T
Apolipoprotein C3	APOC3	T
Apolipoprotein D	APOD	T
Apolipoprotein E	APOE	T
Apolipoprotein H	APOH	T
Apopain	CPP32	T
Apoptosis antigen 1	APT1	G
Apoptosis antigen ligand 1	APT1LG1	I
Apoptosis-inducing factor	AIF	I
Apurinic endonuclease	APE	E
Archaete-scute homolog 1	ASH1	G
Archaete-scute homolog 2	ASH2	G
Arginosuccinate synthetase	ASS	E
Arrestin	SAG	S
Aryl hydrocarbon receptor	AHR	T
Aryl hydrocarbon receptor nuclear translocator	ARNT	T
Arylsulfatase A	ARSA	E
Arylsulfatase B	ARSB	E
Arylsulfatase C	ARSC1	E
Arylsulfatase D	ARSD	E
Arylsulfatase E	ARSE	E
Arylsulfatase F	ARSF	E
Aspartate transaminase		E
Aspartate transcarbamoylase		T
Aspartoacylase	ASPA	E
Aspartylglucosaminidase	AGA	E
Astrotactin	ASTN	E
Ataxia telangiectasia complementation group D	ATD, ATDC	G
Ataxia telangiectasia gene, AT	ATM	G
Ataxin 1	SCA1	G
Ataxin 2	SCA2	G
Ataxin 3	MJD	G
ATP-binding cassette transporter 7	ABC7	I
Atrial natriuretic peptide	ANP	G
Atrial natriuretic peptide receptor A	NPR1	G

Atrial natriuretic peptide receptor B	NPR2	G
Atrial natriuretic peptide receptor C	NPR3	G
Atrophin 1	DRPLA	G
Attractin		I
Autoimmune regulator, AIRE	AIRE	I
Azoospermia factor 1	AZF1	G
Bagpipe homeobox, drosophila homolog of, 1	BAPX1	G
B-cell CLL/lymphoma 1	BCL1	I
B-cell CLL/lymphoma 10	BCL10	I
B-cell CLL/lymphoma 3	BCL3	I
B-cell CLL/lymphoma 4	BCL4	I
B-cell CLL/lymphoma 5	BCL5	I
B-cell CLL/lymphoma 6	BCL6	I
B-cell CLL/lymphoma 7	BCL7	I
B-cell CLL/lymphoma 8	BCL8	I
B-cell CLL/lymphoma 9	BCL9	I
BCL2-associated X protein	BAX	G
BCL2-related protein A1	BCL2A1	G
Beckwith-Wiedemann region 1A	BWR1A	G
Bestrophin	VMD2	T
beta 2 microglobulin	B2M	I
beta-endorphin receptor		N
beta-Glucuronidase	GUSB	E
beta-N-acetylhexosaminidase, A		E
beta-N-acetylhexosaminidase, B		E
Bilirubin UDP-glucuronosyltransferase		E
Bleomycin hydrolase	BLMH	E
Bloom syndrome protein	BLM	G
Blue cone pigment	BCP	S
Bone morphogenetic protein, BMP1	BMP1	G
Bone morphogenetic protein, BMP2	BMP2	G
Bone morphogenetic protein, BMP3	BMP3	G
Bone morphogenetic protein, BMP4	BMP4	G
Bone morphogenetic protein, BMP5	BMP5	G
Bone morphogenetic protein, BMP6	BMP6	G
Bone morphogenetic protein, BMP7	BMP7	G
Bone morphogenetic protein, BMP8	BMP8	G
Brain derived neurotrophic factor	BDNF	G
Brain derived neurotrophic factor (BDNF) receptor	BDNFR	G
Branched chain aminotransferase 1, cytosolic	BCAT1	E
Branched chain aminotransferase 2, mitochondrial	BCAT2	E
BRCA1-associated RING domain gene 1	BARD1	G
Breakpoint cluster region	BCR	G
Breast cancer 1	BRCA1	G
Breast cancer 2	BRCA2	G
Breast cancer, ductal, 1	BRCD1	G

Breast cancer, ductal, 2	BRC2D	G
Bruton agammaglobulinaemia tyrosine kinase	BTX	G
Butyrylcholinesterase	BCHE	E
C3 convertase		E
Ca(2+) transporting ATPase, fast twitch	ATP2A1	T
Ca(2+) transporting ATPase, slow twitch	ATP2A2	T
Cadherin E	CDH1	G
Cadherin EP		G
Cadherin N	CDH2	G
Cadherin P	CDH3	G
Calbindin 1	CALB1	G
Calbindin D9K	CALB3	G
Calcium channel, voltage-dependent, alpha 1F subunit	CACNA1F	N
Calcium channel, voltage-dependent, Alpha-1B (CACNL1A5)	CACNA1B	N
Calcium channel, voltage-dependent, Alpha-1C	CACNA1C	N
Calcium channel, voltage-dependent, Alpha-1D	CACNA1D	N
Calcium channel, voltage-dependent, Alpha-1E (CACNL1A6)	CACNA1E	N
Calcium channel, voltage-dependent, Alpha-2/delta	CACNA2	N
Calcium channel, voltage-dependent, Beta 1	CACNB1	N
Calcium channel, voltage-dependent, Beta 3	CACNB3	N
Calcium channel, voltage-dependent, L type, alpha 1S subunit	CACNA1S	N
Calcium channel, voltage-dependent, Neuronal, Gamma	CACNG2	N
Calcium channel, voltage-dependent, P/Q type, alpha 1A subunit	CACNA1A	N
Calcium channel, voltage-dependent, T-type		N
Calcium sensing receptor	CASR	T
Calmodulin 1	CALM1	G
Calmodulin 2	CALM2	G
Calmodulin 3	CALM3	G
Calmodulin dependant kinase		T
Calmodulin-dependant protein kinase II	CAMK2A	G
Calnexin	CANX	G
Calpain	CAPN, CAPN3	E
Canalicular multispecific organic anion transporter	CMOAT	T
Carbamoylphosphate synthetase 1	CPS1	E
Carbamoylphosphate synthetase 2	CPS2	E
Carbonic anhydrase 3	CA3	E
Carbonic anhydrase 4	CA4	E
Carbonic anhydrase, alpha	CA1	E

Carbonic anhydrase, beta	CA2	E
Cardiac-specific homeobox, CSX	CSX	G
Carnitine acetyltransferase	CRAT	E
Carnitine acylcarnitine translocase	CACT	E
Carnitine transporter protein	CDSP, SCD	T
Cartilage oligomeric matrix protein	COMP, EDM1, PSACH	N
Cartilage-hair hypoplasia gene	CHH	N
Caspase 1	CASP1	G
Caspase 10	CASP10	G
Caspase 2	CASP2	G
Caspase 3	CASP3	G
Caspase 4	CASP4	G
Caspase 5	CASP5	G
Caspase 6	CASP6	G
Caspase 7	CASP7	G
Caspase 8	CASP8	G
Caspase 9	CASP9	G
Catechol-O-methyltransferase	COMT	E
Catenin, alpha	CTNNA1	G
Catenin, beta	CTNNB1	G
Catenin, gamma		G
Cathepsin K	CTSK	E
Caveolin 3	CAV3	E
CD1	CD1	I
CD44	CD44	I
Cdc 25 phosphatase		G
Cdc2	CDC2	G
CDX1		G
CEA		G
Cell adhesion molecule, intercellular, ICAM	ICAM1	G
Cell adhesion molecule, leukocyte-endothelial, LECAM (CD62)	LECAM1	G
Cell adhesion molecule, liver, LCAM	LCAM	G
Cell adhesion molecule, neural, NCAM1	NCAM1	G
Cell adhesion molecule, neural, NCAM120	NCAM120	G
Cell adhesion molecule, neural, NCAM2	NCAM2	G
Cell adhesion molecule, platelet-endothelial, PECAM	PECAM1	G
Cell adhesion molecule, vascular, VCAM	VCAM1	G
Cellubrevin	CEB	N
c-erbB1	ERBB1	G
c-erbB2	ERBB2	G
c-erbB3	ERBB3	G
c-erbB4	ERBB4	G
Ceroid lipofuscinosis neuronal 2	CLN2	N
Ceroid lipofuscinosis neuronal 3	CLN3	N
Ceroid lipofuscinosis neuronal 4	CLN4	N

Ceroid lipofuscinosis neuronal 5	CLN5	N
Ceroid lipofuscinosis neuronal 6	CLN6	N
Chediak-Higashi syndrome 1 gene	CHS1	T
Chemokine MCAF	MCAF	I
Chemokine receptor CCR2	CCR2	I
Chemokine receptor CCR3	CCR3	I
Chemokine receptor CCR5	CCR5	I
Chemokine receptor CXCR1	CXCR1	I
Chemokine receptor CXCR2	CXCR2	I
Chemokine receptor CXCR4	CXCR4	I
Chloride channel 5	CLCN5	S
Cholestasis, progressive familial intrahepatic 1 gene	FIC1	G
Cholesterol ester transfer protein	CETP	T
Choline acetyltransferase	CHAT	E
Choroideremia gene	CHM	S
Chromogranin A	CHGA	G
Ciliary neurotrophic factor (CNTF)	CNTF	G
Ciliary neurotrophic factor (CNTF) receptor	CNTFR	G
c-kit receptor tyrosine kinase		G
Clathrin		T
Cleavage signal-1 protein	CS1	G
Cleft palate gene	CPX	G
Clusterin	CLU	G
CoA transferase		E
Cochlin	COCH	I
Cockayne syndrome gene, CKN1	CKN1	G
Collagen I alpha 1	COL1A1	S
Collagen I alpha 2	COL1A2	S
Collagen II alpha 1	COL2A1	S
Collagen III alpha 1	COL3A1	S
Collagen IV alpha 1	COL4A1	S
Collagen IV alpha 2	COL4A2	S
Collagen IV alpha 3	COL4A3	S
Collagen IV alpha 4	COL4A4	S
Collagen IV alpha 5	COL4A5	S
Collagen IV alpha 6	COL4A6	S
Collagen IX alpha 2	COL9A2, EDM2	S
Collagen IX alpha 3	COL9A3	S
Collagen receptor	COLR	S
Collagen V alpha 1	COL5A1	S
Collagen V alpha 2	COL5A2	S
Collagen VI alpha 1	COL6A1	S
Collagen VI alpha 2	COL6A2	S
Collagen VI alpha 3	COL6A3	S
Collagen VII alpha 1	COL7A1	S
Collagen X alpha 1	COL10A1	S
Collagen X alpha 1	COL11A1	S

Collagen XI alpha 2	COL11A2	S
Collagen XVII alpha 1	COL17A1	S
Collagenic-like tail subunit of asymmetric acetylcholinesterase	COLQ	E
Collapsin		G
Colony-stimulating factor 1	CSF1	G
Colony-stimulating factor 1 receptor	CSF1R	G
Colony-stimulating factor 2	CSF2	G
Colony-stimulating factor 2 alpha receptor	CSF2RA	G
Colony-stimulating factor 2 beta receptor	CSF2RB	G
Colony-stimulating factor 3	CSF3	G
Colony-stimulating factor 3 receptor	CSF3R	G
Complex V	MTATP6	E
Cone-rod homeobox-containing gene	CRX	G
Contactin	CNTN1	G
Core-binding factor, alpha 1	CBFA1	G
Core-binding factor, alpha 2	CBFA2	G
Core-binding factor, beta	CBFB	G
Corticotrophin-releasing hormone	CRH	T
Corticotrophin-releasing hormone receptor	CRHR1	T
Creatine kinase - B and m	CKBE	E
Creb binding protein	CREBBP	G
Cryptochrome 1	CRY1	S
Cryptochrome 2	CRY2	S
Crystallin, alpha A	CRYAA	S
Crystallin, alpha B	CRYAB	S
Crystallin, beta B2	CRYBB2	S
Crystallin, gamma A	CRYGA	S
c-src tyrosine kinase	CSK	G
Cu ²⁺ transporting ATPase alpha polypeptide	ATP7A	E
Cu ²⁺ transporting ATPase beta polypeptide	ATP7B	E
Cubilin	CUBN	T
Cyclic AMP response element binding protein	CREB	G
Cyclic AMP response element modulator	CREM	G
Cyclic AMP-dependent protein kinase	PKA	E
Cyclic nucleotide gated channel alpha 1, CNGA1	CNGA1	N
Cyclic nucleotide gated channel alpha 3, CNGA3	CNGA3	N
Cyclic nucleotide phosphodiesterase 1B	PDE1B	E
Cyclic nucleotide phosphodiesterase 1B1	PDE1B1	E
Cyclic nucleotide phosphodiesterase 2A3	PDE2A3	E
Cyclic nucleotide phosphodiesterase 3A	PDE3A	E
Cyclic nucleotide phosphodiesterase 3B	PDE3B	E
Cyclic nucleotide phosphodiesterase 4A	PDE4A	E
Cyclic nucleotide phosphodiesterase 4C	PDE4C	E
Cyclic nucleotide phosphodiesterase 5A	PDE5A	E
Cyclic nucleotide phosphodiesterase 6A	PDE6A	E

Cyclic nucleotide phosphodiesterase 6B	PDE6B	E
Cyclic nucleotide phosphodiesterase 7	PDE7	E
Cyclic nucleotide phosphodiesterase 8	PDE8	E
Cyclic nucleotide phosphodiesterase 9A	PDE9A	E
Cyclin A	CCNA	G
Cyclin B	CCNB	G
Cyclin C	CCNC	G
Cyclin D	CCND1	G
Cyclin E	CCNE	G
Cyclin F	CCNF	G
Cyclin-dependent kinase 1	CDK1	G
Cyclin-dependent kinase 10	CDK10	G
Cyclin-dependent kinase 2	CDK2	G
Cyclin-dependent kinase 3	CDK3	G
Cyclin-dependent kinase 4	CDK4	G
Cyclin-dependent kinase 5	CDK5	G
Cyclin-dependent kinase 6	CDK6	G
Cyclin-dependent kinase 7	CDK7	G
Cyclin-dependent kinase 8	CDK8	G
Cyclin-dependent kinase 9	CDK9	G
Cyclin-dependent kinase inhibitor 1A (P21, CIP1)	CDKN1A	G
Cyclin-dependent kinase inhibitor 1B (P27, KIP1)	CDKN1B	G
Cyclin-dependent kinase inhibitor 1C (P57, KIP2)	CDKN1C	G
Cyclin-dependent kinase inhibitor 2A (p16)	CDKN2A	G
Cyclin-dependent kinase inhibitor 3	CDKN3	G
Cyclooxygenase 1	COX1	E
Cyclooxygenase 2	COX2	E
CYP11A1	CYP11A1	E
CYP11B1	CYP11B1	E
CYP11B2	CYP11B2	E
CYP17	CYP17	E
CYP19	CYP19	E
CYP1A1	CYP1A1	E
CYP1A2	CYP1A2	E
CYP1B1	CYP1B1	E
CYP21	CYP21	E
CYP24	CYP24	E
CYP27	CYP27	E
CYP27B1	PDDR	E
CYP2A1	CYP2A1	E
CYP2A13	CYP2A13	E
CYP2A3	CYP2A3	E
CYP2A6V2	CYP2A6V2	E
CYP2A7	CYP2A7	E
CYP2B6	CYP2B6	E

CYP2C18	CYP2C18	E
CYP2C19	CYP2C19	E
CYP2C8	CYP2C8	E
CYP2C9	CYP2C9	E
CYP2D6	CYP2D6	E
CYP2E1	CYP2E1	E
CYP2F1	CYP2F1	E
CYP2J2	CYP2J2	E
CYP3A3	CYP3A3	E
CYP3A4	CYP3A4	E
CYP3A5	CYP3A5	E
CYP3A7	CYP3A7	E
CYP4A11	CYP4A11	E
CYP4B1	CYP4B1	E
CYP4F2	CYP4F2	E
CYP4F3	CYP4F3	E
CYP51	CYP51	E
CYP5A1	CYP5A1	E
CYP7A	CYP7A	E
CYP8	CYP8	E
Cystathionase	CTH	E
Cystathione beta synthase	CBS	E
Cystic fibrosis transmembrane conductance regulator, CFTR	CFTR	N
Cystinosin	CTNS	T
Cytidine deaminase	CDA	E
Cytochrome b-245 alpha	CYBA	E
Cytochrome b-245 beta	CYBB	E
Cytochrome b-5	CYB5	E
DAX1 nuclear receptor	DAX1	I
Deafness autosomal dominant 5	DFNA5	N
Deafness dystonia peptide	DDP	N
Defender against cell death 1	DAD1	G
Deleted in azoospermia	DAZ	G
Deleted in colorectal carcinoma	DCC	G
Deleted in malignant brain tumours 1	DMBT1	G
Delta aminolevulinate dehydratase	ALAD	E
Delta(4)-3-oxosteroid 5-beta-reductase		E
Delta-7-dehydrocholesterol reductase	DHCR7	E
Dentin sialophosphoprotein	DSPP	G
Deoxyuridine triphosphatase; dUTPase		E
Desert hedgehog, dhh		G
DHEA sulfotransferase	STD	E
Diaphanous 1	DIAPH1	N
Diaphanous 2	DIAPH2	N
Diastrophic dysplasia sulfate transporter	DTD	T
Dihydrolipoamide branched chain transacylase	DBT	N
Dihydrolipoamide dehydrogenase	DLD	N

Dihydrolipoyl dehydrogenase 2	PDHA	E
Dihydrolipoyl transacetylase	PDHA	E
Dihydroorotase		E
Dihydroxyacetonephosphate acyltransferase	DHAPAT	E
Disrupted meiotic cDNA 1, homolog	DMC1	G
Distal-less homeobox 1	DLX1	G
Distal-less homeobox 2	DLX2	G
Distal-less homeobox 3	DLX3	G
Distal-less homeobox 4	DLX4	G
Distal-less homeobox 5	DLX5	G
Distal-less homeobox 6	DLX6	G
DNA damage binding protein, DDB1	DDB1	S
DNA damage binding protein, DDB2	DDB2	S
DNA directed polymerase, alpha	POLA	E
DNA glycosylases		E
DNA helicases		E
DNA Ligase 1	LIG1	E
DNA methyltransferase	DNMT	E
DNA polymerase 1		E
DNA polymerase 2		E
DNA polymerase 3		E
DNA primase		E
DNA-damage-inducible transcript 3	DDIT3	S
DNA-dependant RNA polymerase		E
DOPA decarboxylase	DDC	E
Doublecortin, DCX	DCX	S
Duffy blood group	FY	T
Dynamin	DNM1	G
Dynein		G
Dyskerin	DKC1	S
Dystonia 1	DYT1	S
Dystonia 3	DYT3	S
Dystonia 6	DYT6	S
Dystonia 7	DYT7	S
Dystonia 9	CSE	S
Dystrophia myotonica	DM, DMPK	E
Dystrophia myotonica, atypical	DM2	E
Dystrophin	DMD	S
Dystrophin-associated glycoprotein 35kD, SCGD	SGCD	S
Dystrophin-associated glycoprotein 35kD, SGSG	SGCG	S
Dystrophin-associated glycoprotein 43kD	SGCB	S
Dystrophin-associated glycoprotein 50kD	SGCA	S
E74-like factor 1, ELF1	ELF1	G
EB1		G
Ectodermal Dysplasia 1 gene	ED1	S
Electron-transferring-flavoprotein alpha	ETFA	T

Electron-transferring-flavoprotein beta	ETFB	T
Electron-transferring flavoprotein dehydrogenase	ETFDH	E
Empty spiracles (drosophila) homologue 1	EMX1	G
Empty spiracles (drosophila) homologue 2	EMX2	G
Endobrevin	VAMP8	N
Endocardial fibroelastosis 2 gene	EFE2	S
Endometrial bleeding-associated factor	EBAF	G
Endothelin 1	EDN1	N
Endothelin 2	EDN2	N
Endothelin 3	EDN3	N
Endothelin converting enzyme	ECE1	N
Endothelin receptor type A	EDNRA	N
Endothelin receptor type B	EDNRB	N
Engrailed-1	EN1	G
Engrailed-2	EN2	G
Enolase	ENO1	E
Enoyl CoA isomerase		E
Enterokinase	PRSS7, ENTK	E
Ephrin receptor tyrosine kinase A	EPHA	G
Ephrin receptor tyrosine kinase B	EPHB	G
Ephrin-A	EFNA	G
Ephrin-B	EFNB	G
Epidermal growth factor	EGF	G
Epidermal growth factor receptor	EGFR	G
Epilepsy, benign neonatal 4 gene	ICCA	E
Epilepsy, female restricted	EFMR	E
Epilepsy, progressive myoclonic 2 gene	EPM2A	E
Erythrocyte membrane protein band 4.1	EPB41	S
Erythrocyte membrane protein band 4.2	EPB42	S
Erythrocyte membrane protein band 7.2	EPB72	S
Erythroid kruppel-like factor	EKLF	G
Erythropoietin	EPO	I
Erythropoietin receptor	EPOR	I
Estrogen receptor	ESR	G
Eukaryotic initiation translation factor	EIF4E	G
EWS RNA-binding protein	EWSR1	G
Excision repair complementation group 1 protein	ERCC1	E
Excision repair complementation group 2 protein	ERCC2	E
Excision repair complementation group 2 protein	ERCC3	E
Excision repair complementation group 4 protein	ERCC4	E
Excision repair complementation group 6 protein	ERCC6	E
Exostosin 1	EXT1	S

Exostosin 2	EXT2	S
Exostosin 3	EXT3	S
Eyes absent 1	EYA1	G
Eyes absent 2	EYA2	G
Eyes absent 3	EYA3	G
Faciogenital dysplasia	FGD1, FGDY	T
Factor 1 (No. one)	F1	I
Factor B, properdin		I
Factor D		I
Factor H	HF1	I
Factor I (letter I)	IF	I
Factor III	F3	I
Factor IX	F9	I
Factor V	F5	I
Factor VII	F7	I
Factor VIII	F8	I
Factor X	F10	I
Factor XI	F11	I
Factor XII	F12	I
Factor XIII A & B	F13A & F13B	I
Fanconi anemia, complementation group A	FANCA	T
Fanconi anemia, complementation group C	FANCC	T
Fanconi anemia, complementation group D	FANCD	T
Fc fragment of IgG, high affinity IA, receptor for	FCGR1A	G
Fc fragment of IgG, low affinity IIa, receptor for	FCGR2A	G
(CD32)		
Fc fragment of IgG, low affinity IIIa, receptor for	FCGR3A	G
(CD16)		
Fc receptor		I
Fertilin protein	FTNB	G
Fibrillin 1	FBN1	G
Fibrillin 2	FBN2	G
Fibroblast growth factor	FGF1	G
Fibroblast growth factor receptor 1	FGFR1	G
Fibroblast growth factor receptor 2	FGFR2	G
Fibroblast growth factor receptor 3	FGFR3	G
Fibronectin precursor	FN1	G
Flavin-containing monooxygenase 1	FMO1	E
Flavin-containing monooxygenase 2	FMO2	E
Flavin-containing monooxygenase 3	FMO3	E
Flavin-containing monooxygenase 4	FMO4	E
Flightless-II, Drosophila homolog of	FLII	G
Folic acid receptor	FOLR	G
Follicle stimulating hormone receptor	FSHR, ODG1	G
Follicle stimulating hormone, FSH	FSHB	G
Follicular lymphoma variant translocation 1	FVT1	I
Follistatin		G
Forkhead rhabdomyosarcoma gene	FKHR	G

Forkhead transcription factor 10	FKHL10	G
Forkhead transcription factor 14	FKHL14	G
Forkhead transcription factor 7	FKHL7	G
Formiminotransferase		E
Fragile site, folic acid type, rare, fra(X) A	FRAXA	N
Fragile site, folic acid type, rare, fra(X) E	FRAXE	N
Fragile site, folic acid type, rare, fra(X) F	FRAXF	N
Frataxin	FRDA	G
Fringe secreted protein, lunatic	LFNG	G
Fringe secreted protein, manic	MFNG	G
Fringe secreted protein, radical	RFNG	G
Fructose-1,6-diphosphatase	FBP1	E
Fucosyltransferase 6	FUT6	T
Fukuyama type congenital muscular dystrophy	FCMD	G
Fumarase	FH	E
Fumarylacetoacetase	FAH	E
G/T mismatch binding protein	GTBP, MSH6	G
GABA receptor, alpha 1	GABRA1	N
GABA receptor, alpha 2	GABRA2	N
GABA receptor, alpha 3	GABRA3	N
GABA receptor, alpha 4	GABRA4	N
GABA receptor, alpha 5	GABRA5	N
GABA receptor, alpha 6	GABRA6	N
GABA receptor, beta 1	GABRB1	N
GABA receptor, beta 2	GABRB2	N
GABA receptor, beta 3	GABRB3	N
GABA receptor, gamma 1	GABRG1	N
GABA receptor, gamma 2	GABRG2	N
GABA receptor, gamma 3	GABRG3	N
GABA transaminase	ABAT	E
Gadd45 (growth arrest & DNA-damage-inducible protein)		E
Galactocerebrosidase	GALC	E
Galactokinase	GALK1	E
Galactose 1-phosphate uridyl-transferase	GALT	E
Galactosyltransferase 1	GT1	G
Galactosyltransferase, alpha 1,3	GGTA1	G
Galactosyltransferase, beta 3	B3GALT	G
Galanin	GAL	N
Galanin receptor	GALNR1	N
Gamma-glutamyl carboxylase	GGCX	T
Gap junction protein alpha 1	GJA1	T
Gap junction protein alpha 3	GJA3	T
Gap junction protein alpha 8	GJA8	T
Gap junction protein beta 1	GJB1	T
Gap junction protein beta 2	GJB2	T
Gap junction protein beta 3	GJB3	T
Gastric Intrinsic factor, GIF	GIF	E
Gastrin	GAS	G

Gastrin releasing peptide	GRP	T
Gastrointestinal tumor-associated antigen 1	GA733	I
Gastrulation brain homeobox 2	GBX2	G
GDP dissociation inhibitor 1	GDI1	G
Gelsolin	GSN	G
Geniospasm 1	GSM1	G
Gephyrin		N
Glial-cell derived neurotrophic factor (GDNF) receptor		N
Glial-cell derived neurotrophic factor, GDNF	GDNF	N
Glioma chloride ion channel, GCC		G
Glucagon receptor	GCGR	G
Glucagon-like peptide receptor 1	GLP1R	G
Glucocorticoid receptor	GRL	G
Glucose-6-phosphatase translocase	G6PT1	E
Glucosidase, acid alpha	GAA	E
Glucosidase, acid beta	GBA	E
Glutamate decarboxylase, GAD	GAD1	E
Glutamate-cysteine ligase	GLCLC	E
Glutathione	GSH	T
Glutathione peroxidase, GPX1	GPX1	E
Glutathione peroxidase, GPX2	GPX2	E
Glutathione reductase, GSR	GSR	E
Glutathione S-transferase mu 1, GSTM1	GSTM1	E
Glutathione S-transferase mu 4, GSTM4		E
Glutathione S-transferase theta 1, GSTT1	GSTT1	E
Glutathione S-transferase theta 2, GSTT2		E
Glutathione S-transferase, GSTP1	GSTP1	E
Glutathione S-transferase, GSTZ1	GSTZ1	E
Glutathione synthetase	GSS	E
Glyceraldehyde-3-phosphate dehydrogenase, GAPDH	GAPDH	E
Glycerol kinase	GK	E
Glycinamide ribonucleotide (GAR) transformylase	GART	E
Glycine dehydrogenase	GLDC	E
Glycine receptor, alpha	GLRA2	N
Glycine receptor, beta		N
Glycogen branching enzyme	GBE1	E
Glycogen phosphorylase	PYGL	E
Glycogen synthase 1 (muscle)	GLYS1	E
Glycogen synthase 2 (liver)	GYS2	E
Glycosyltransferases, ABO blood group	ABO	E
Glypican 3	GPC3, SDYS	G
GM2 ganglioside activator protein, GM2A	GM2A	E
Gonadotropin releasing hormone	GNRH	G
Gonadotropin releasing hormone receptor	GNRHR	G
Goosecoid GSC		G

Green cone pigment	GCP	S
Growth arrest-specific homeobox	GAX	G
Growth factor receptor-bound protein 2	GRB2	G
Growth hormone 1	GH1	G
Growth hormone 2 (placental)	GH2	G
Growth hormone receptor	GHR	G
Growth hormone releasing hormone (GHRH)	GHRH	G
Growth hormone releasing hormone receptor	GHRHR	G
Growth/differentiation factor 5	GDF5	G
Growth-regulated protein precursor, GRO	GRO	I
GTP cylcohydrolase 1	GCH1	G
GTPase-activating protein, GAP	RASA1	G
Guanidinoacetate N-methyltransferase	GAMT	E
Guanine nucleotide-binding protein, alpha activating activity polypeptide, GNAO	GNAO1	N
Guanine nucleotide-binding protein, alpha inhibiting activity polypeptide 1, GNAI1	GNAI1	N
Guanine nucleotide-binding protein, alpha inhibiting activity polypeptide 2, GNAI2	GNAI2	N
Guanine nucleotide-binding protein, alpha inhibiting activity polypeptide 3, GNAI3	GNAI3	N
Guanine nucleotide-binding protein, alpha stimulating activity polypeptide, GNAS1	GNAS1	N
Guanine nucleotide-binding protein, alpha stimulating activity polypeptide, GNAS2	GNAS2	N
Guanine nucleotide-binding protein, alpha stimulating activity polypeptide, GNAS3	GNAS3	N
Guanine nucleotide-binding protein, alpha stimulating activity polypeptide, GNAS4	GNAS4	N
Guanine nucleotide-binding protein, alpha transducing activity polypeptide, GNAT1	GNAT1	N
Guanine nucleotide-binding protein, alpha transducing activity polypeptide, GNAT2	GNAT2	N
Guanine nucleotide-binding protein, beta polypeptide 3	GNB3	N
Guanine nucleotide-binding protein, gamma polypeptide 5	GNG5	N
Guanine nucleotide-binding protein, q polypeptide	GNAQ	N
Guanylate cyclase 2D, membrane (retina-specific)	GUCY2D	E
Guanylate cyclase activator 1A (retina)	GUCA1A	E
Guanylate kinase		E
Gustducin, alpha (taste-specific G protein)	GDCA	N
Haeme regulated inhibitor kinase		E
Haemoglobin epsilon		T
Hairless	HR	G
Haptoglobin, alpha 1	HPA1	I

Haptoglobin, alpha 2	HPA2	I
Haptoglobin, beta	HPB	I
Heat shock protein, HSP60		I
Heat shock protein, HSP70		I
Heat shock protein, HSP90		I
Heat shock protein, HSPA1		I
Heat shock protein, HSPA2		I
Hela tumor suppression gene	HTS1	G
Hemochromatosis	HFE	T
Hemopexin	HPX	I
Heparan sulfamidase		E
Heparin binding epidermal growth factor	HBEGF	G
Hepatic nuclear factor-3-beta	HNF3B	E
Hepatic nuclear factor-4-alpha	HNF4A	E
Hepatitis B virus integration site 1	HVBS1	I
Hepatitis B virus integration site 2	HVBS6	I
Hepatocyte growth factor	HGF	G
Hexosaminidase A	HEXA,TSD	E
Hexosaminidase B	HEXB	E
High mobility group protein 1	HMG1	G
High mobility group protein 2	HMG2	G
High mobility group protein C	HMGIC	G
High mobility group protein Y	HMG1Y	G
Histone family H1	H1	G
Histone family H2	H2	G
Histone family H3	H3	G
Histone family H4	H4	G
HLA-B associated transcript 1	BAT1	I
HLH transcription factor HAND1	HAND1	G
HLH transcription factor HAND2	HAND2	G
HMG-CoA lyase	HMGCL	E
HMG-CoA reductase	HMGCR	E
HMG-CoA synthase	HMGCS2	E
Holocarboxylase synthetase	HLCS	E
Holoprosencephaly 1	HPE1	G
Holoprosencephaly 2	HPE2	G
Holoprosencephaly 3	HPE3	G
Holoprosencephaly 4	HPE4	G
Homeobox (HOX) gene A1	HOXA1	G
Homeobox (HOX) gene A10	HOXA10	G
Homeobox (HOX) gene A11	HOXA11	G
Homeobox (HOX) gene A12	HOXA12	G
Homeobox (HOX) gene A13	HOXA13	G
Homeobox (HOX) gene A2	HOXA2	G
Homeobox (HOX) gene A3	HOXA3	G
Homeobox (HOX) gene A4	HOXA4	G
Homeobox (HOX) gene A5	HOXA5	G
Homeobox (HOX) gene A6	HOXA6	G

Homeobox (HOX) gene A7	HOXA7	G
Homeobox (HOX) gene A8	HOXA8	G
Homeobox (HOX) gene A9	HOXA9	G
Homeobox (HOX) gene B1	HOXB1	G
Homeobox (HOX) gene B2	HOXB2	G
Homeobox (HOX) gene B3	HOXB3	G
Homeobox (HOX) gene B4	HOXB4	G
Homeobox (HOX) gene B5	HOXB5	G
Homeobox (HOX) gene B6	HOXB6	G
Homeobox (HOX) gene B7	HOXB7	G
Homeobox (HOX) gene B8	HOXB8	G
Homeobox (HOX) gene B9	HOXB9	G
Homeobox (HOX) gene C13	HOXC13	G
Homeobox (HOX) gene C4	HOXC4	G
Homeobox (HOX) gene C8	HOXC8	G
Homeobox (HOX) gene C9	HOXC9	G
Homeobox (HOX) gene D1	HOXD1	G
Homeobox (HOX) gene D10	HOXD10	G
Homeobox (HOX) gene D12	HOXD12	G
Homeobox (HOX) gene D13	HOXD13	G
Homeobox (HOX) gene D3	HOXD3	G
Homeobox (HOX) gene D4	HOXD4	G
Homeobox (HOX) gene D8	HOXD8	G
Homeobox (HOX) gene D9	HOXD9	G
Homeobox 11	HOX11	G
Homeobox HB24	HLX1	G
Homeobox HB9	HLXB9	G
Homeobox, PROX1	PROX1	G
HSSB, replication protein		E
Human atonal gene	ATOH1	G
Human chorionic gonadotrophin, hCG	CG	G
Human placental lactogen	CSH1	G
Huntingtin	HD	T
Hypoxanthine-guanine phosphoribosyltransferase, HGPRT	HPRT	E
Hypoxia inducible factor 1	HIF1A	E
Hypoxia inducible factor 2		E
IC7 A and B		I
Iduronate 2 sulphatase	IDS	E
Ikaros gene	IKAROS	G
Immunoglobulin alpha (IgA)	IGHA	I
Immunoglobulin delta (IgD)	IGHD	I
Immunoglobulin E (IgE) responsiveness gene	IGER	I
Immunoglobulin E (IgE) serum concentration regulator gene	IGES	I
Immunoglobulin epsilon (IgE)	IGHE	I
Immunoglobulin gamma (IgG) 2	IGHG2	I
Immunoglobulin heavy mu chain	IGHM	I

Immunoglobulin J polypeptide	IGJ	I
Immunoglobulin kappa constant region	IGKC	I
Immunoglobulin kappa variable region	IGKV	I
Indian hedgehog, ihh	IHH	G
Inhibin, alpha	INHA	G
Inhibin, beta A	INHBA	G
Inhibin, beta B	INHBB	G
Inhibin, beta C	INHBC	G
Inosine monophosphate dehydrogenase, IMPDH		E
Inositol 1,4,5-triphosphate receptor 1	ITPR1	G
Inositol 1,4,5-triphosphate receptor 3	ITPR3	G
Insulin	INS	G
Insulin promotor factor 1	IPF1	G
Insulin receptor	INSR	G
Insulin receptor substrate-1	IRS1	G
Insulin-like growth factor 1	IGF1	G
Insulin-like growth factor 1 receptor	IGF1R	G
Insulin-like growth factor 2	IGF2	G
Insulin-like growth factor 2 receptor	IGF2R	G
Integrin beta 1	ITGB1	G
Integrin beta 2	ITGB2	G
Integrin beta 3	ITGB3	G
Integrin beta 4	ITGB4	G
Integrin beta 5	ITGB5	G
Integrin beta 6	ITGB6	G
Integrin beta 7	ITGB7	G
Integrin, alpha 1	ITGA1	G
Integrin, alpha 2	ITGA2	G
Integrin, alpha 3	ITGA3	G
Integrin, alpha 4	ITGA4	G
Integrin, alpha 5	ITGA5	G
Integrin, alpha 6	ITGA6	G
Integrin, alpha 7	ITGA7	G
Integrin, alpha 8	ITGA8	G
Integrin, alpha 9	ITGA9	G
Integrin, alpha M	ITGAM	G
Integrin, alpha X	ITGAX	G
Inter-alpha-trypsin inhibitor, IATI		E
Intercellular adhesion molecule 1	ICAM1	I
Intercellular adhesion molecule 2	ICAM2	I
Intercellular adhesion molecule 3	ICAM3	I
Interferon alpha	IFNA1	I
Interferon beta	IFNB	I
Interferon gamma	IFNG	I
Interferon gamma receptor 1	IFNGR1	I
Interferon gamma receptor 2	IFNGR2	I
Interferon regulatory factor 1	IRF1	I

Interferon regulatory factor 4	IRF4	I
Interleukin(IL) 1 receptor	IL1R	I
Interleukin(IL) 1, alpha	IL1A	I
Interleukin(IL) 1, beta	IL1B	I
Interleukin(IL) 10	IL10	I
Interleukin(IL) 10 receptor	IL10R	I
Interleukin(IL) 11	IL11	I
Interleukin(IL) 11 receptor	IL11R	I
Interleukin(IL) 12	IL12	I
Interleukin(IL) 12 receptor, beta 1	IL12RB1	I
Interleukin(IL) 13	IL13	I
Interleukin(IL) 13 receptor	IL13R	I
Interleukin(IL) 2	IL2	I
Interleukin(IL) 2 receptor, alpha	IL2RA	I
Interleukin(IL) 2 receptor, gamma	IL2RG	I
Interleukin(IL) 3	IL3	I
Interleukin(IL) 3 receptor	IL3R	I
Interleukin(IL) 4	IL4	I
Interleukin(IL) 4 receptor	IL4R	I
Interleukin(IL) 5	IL5	I
Interleukin(IL) 5 receptor	IL5R	I
Interleukin(IL) 6	IL6	I
Interleukin(IL) 6 receptor	IL6R	I
Interleukin(IL) 7	IL7	I
Interleukin(IL) 7 receptor	IL7R	I
Interleukin(IL) 8	IL8	I
Interleukin(IL) 8 receptor	IL8R	I
Interleukin(IL) 9	IL9	I
Interleukin(IL) 9 receptor	IL9R	I
Interleukin(IL) receptor antagonist 1	IL1RN, IL1RA	I
IP3 kinase		E
Isocitrate dehydrogenase		E
Isovaleric acid CoA dehydrogenase	IVD	E
Janus kinase 1	JAK1	G
Janus kinase 2	JAK2	G
Janus kinase 3	JAK3	G
Kallman syndrome gene 1	KAL1	G
Kell blood group precursor	XK, KEL	T
Keratin 1	KRT1	S
Keratin 10	KRT10	S
Keratin 11	KRT11	S
Keratin 12	KRT12	S
Keratin 13	KRT13	S
Keratin 14	KRT14	S
Keratin 15	KRT15	S
Keratin 16	KRT16	S
Keratin 17	KRT17,PCHC1	S
Keratin 18	KRT18	S

Keratin 2	KRT2	S
Keratin 3	KRT3	S
Keratin 4	KRT4	S
Keratin 5	KRT5	S
Keratin 6	KRT6	S
Keratin 7	KRT7	S
Keratin 8	KRT8	S
Keratin 9	KRT9	S
Ketohexokinase	KHK	E
Kinectin	KTN1	G
Kinesin, heavy chain	KNSL1	G
Kinesin, light chain	KNS2	G
L1 cell adhesion molecule	L1CAM	N
Lactotransferrin	LTF	T
Lamin A/C	LMNA	G
Laminin 5, alpha 3	LAMA3	G
Laminin 5, beta 3	LAMB3	G
Laminin 5, gamma 2	LAMC2	G
Laminin M	LAMM	G
Laminin receptor 1	LAMR1	G
Latent transforming growth factor-beta binding protein 2	LTBP2	G
Leptin	LEP	G
Leptin receptor	LEPR	G
Leukaemia inhibitory factor	LIF	G
Leukaemia inhibitory factor receptor	LIFR	G
Leukin		I
Leukocyte-specific transcript 1	LST-1	I
Leukotriene A4 hydrolase		I
Leukotriene A4 synthase	LTA4S	E
Leukotriene B4 receptor		I
Leukotriene B4 synthase	LTB4S	E
Leukotriene C4 receptor		I
Leukotriene C4 synthase	LTC4S	E
Leukotriene D4/E4 receptor		I
LH/choriogonadotropin (CG) receptor	LHCGR	G
LIM homeobox protein 1	LHX1	G
LIM homeobox protein 2	LHX2	G
LIM homeobox protein 3	LHX3	G
LIM homeobox protein 4	LHX4	G
LIM homeobox transcription factor 1, beta	LMX1B	G
Limb girdle muscular dystrophy 1A	LGMD1A	G
Limb girdle muscular dystrophy 1B	LGMD1B	G
Limb girdle muscular dystrophy 2G	LGMD2G	G
Limb girdle muscular dystrophy 2H	LGMD2H	G
Limbic associated membrane protein	LAMP	G
LIM-domain only protein 1	LMO1	G
LIM-domain only protein 2	LMO2	G

LIM-domain only protein 3	LMO3	G
LIM-domain only protein 4	LMO4	G
Lipoma-preferred partner gene	LPP	G
Lipoprotein receptor, Low Density	LDLR	T
Lipoxygenase 12 (platelets)	LOG12	I
Lipoxygenase 5 (leukocytes)		I
Long QT-type 2 potassium channels	LQT2, KCNH2	T
Loricrin	LOR	S
Low density lipoprotein receptor-related protein precursor	LRP	T
Luteinizing hormone, beta chain	LHB	G
Lymphoblastic leukemia derived sequence 1	LYL1	I
Lymphocyte-specific protein tyrosine kinase	LCK	I
Lymphoid enhancer-binding factor	LEF-1	G
Lysosome-associated membrane protein 1	LAMP1	G
Lysosome-associated membrane protein 2	LAMP2	G
MAD (mothers against decapentaplegic, Drosophila) homologue 2	MADH2	G
MAD (mothers against decapentaplegic, Drosophila) homologue 3	MADH3	G
MAD (mothers against decapentaplegic, Drosophila) homologue 4	MADH4	G
MADS box transcription-enhancer factor 2A	MEF2A	G
MADS box transcription-enhancer factor 2B	MEF2B	G
MADS box transcription-enhancer factor 2C	MEF2C	G
MADS box transcription-enhancer factor 2D	MEF2D	G
Malate dehydrogenase, mitochondrial	MDH2	E
Malignant proliferation, eosinophil gene	MPE	I
Malonyl CoA decarboxylase		E
Malonyl CoA transferase		E
Mannosidase, alpha B lysosomal	MANB	E
Mannosidase, beta A lysosomal	MANBA	E
MAPK kinase 1	MAPKK1; MEK1	G
MAPK kinase 4	MAPKK4; MEK4; SERK1	G
MAPK kinase 6	MAPKK6; MEK6	G
MAPKK kinase	MAPKKK	G
Matrix Gla protein	MGP	G
Matrix metalloproteinase 1	MMP1	E
Matrix metalloproteinase 10	MMP10	E
Matrix metalloproteinase 11	MMP11	E
Matrix metalloproteinase 12	MMP12	E
Matrix metalloproteinase 13	MMP13	E
Matrix metalloproteinase 14	MMP14	E
Matrix metalloproteinase 15	MMP15	E
Matrix metalloproteinase 16	MMP16	E
Matrix metalloproteinase 17	MMP17	E
Matrix metalloproteinase 18	MMP18	E

Matrix metalloproteinase 19	MMP19	E
Matrix metalloproteinase 2	MMP2	E
Matrix metalloproteinase 3	MMP3, STMY1	E
Matrix metalloproteinase 4	MMP4	E
Matrix metalloproteinase 5	MMP5	E
Matrix metalloproteinase 6	MMP6	E
Matrix metalloproteinase 7	MMP7	E
Matrix metalloproteinase 8	MMP8	E
Matrix metalloproteinase 9	MMP9	E
MAX-interacting protein 1	MXI1	G
MEK kinase, MEKK		E
Melanocortin 1 receptor	MC1R	T
Melanocortin 2 receptor	MC2R	T
Melanocortin 4 receptor	MC4R	T
Menin	MEN1	G
Mesoderm-specific transcript	MEST	G
Methionine adenosyltransferase	MAT1A, MAT2A	E
Methionine synthase	MTR	E
Methionine synthase reductase	MTRR	E
Methylguanine-DNA methyltransferase	MGMT	E
Methylmalonyl-CoA mutase	MUT	E
Mevalonate kinase	MVK	E
MHC Class I: A		I
MHC Class I: B		I
MHC Class I: C		I
MHC Class I: LMP-2, LMP-7		I
MHC Class I: Tap1	ABCR, TAP1	I
MHC Class II: DP	HLA-DPB1	I
MHC Class II: DQ		I
MHC Class II: DR		I
MHC Class II: Tap2	TAP2, PSF2	I
MHC Class II:Complementation group A	MHC2TA	I
MHC Class II:Complementation group B	rfxank	I
MHC Class II:Complementation group C	RFX5	I
MHC Class II:Complementation group D	RFXAP	I
Microphthalmia-associated transcription factor	MITF	G
Microsomal triglyceride transfer protein	MTP	T
Microtubule associated protein	MAP	S
Midline 1	MID1	G
Mismatch repair gene, PMSL1	PMS1	G
Mismatch repair gene, PMSL2	PMS2	G
Mitochondrial trifunctional protein, alpha subunit	HADHA	E
Mitochondrial trifunctional protein, beta subunit	HADHB	E
Mitogen-activated protein (MAP) kinase	MAPK	G
Molybdenum cofactor synthesis 1	MOCS1	E
Molybdenum cofactor synthesis 2	MOCS2	E
Monoamine oxidase A	MAOA	E

Monoamine oxidase B	MAOB	E
Monocyte chemoattractant protein 1	MCP1	I
Motilin	MLN	G
Msh homeobox homolog 1	MSX1	G
Msh homeobox homolog 2	MSX2	G
Mucopolidoses	GNPTA	E
Mulibrey nanism	MUL	T
Multidrug resistance associated protein	MRP	G
Muscarinic receptor, M1	CHRM1	N
Muscarinic receptor, M2	CHRM2	N
Muscarinic receptor, M3	CHRM3	N
Muscarinic receptor, M4	CHRM4	N
Muscarinic receptor, M5	CHRM5	N
Muscle phosphorylase	PYGM	E
Mutated in colorectal cancers, MCC	MCC	G
MutL homolog 1	MLH1	G
MutS homolog 2	MSH2	G
MutS homolog 3	MSH3	G
Myelin protein peripheral 22	PMP22	S
Myelin protein zero	MPZ	S
Myelodysplasia syndrome 1 gene	MDS1	G
Myeloid leukemia factor-1	MLF1	I
Myocilin	MYOC	T
Myogenic factor 3	MYF3	G
Myogenic factor 4	MYF4	G
Myogenic factor 5	MYF5	G
Myomesin 1	MYOM1	S
Myomesin 2	MYOM2	S
Myosin 15	MYO15	S
Myosin 6	MYO6	S
Myosin 7A	MYO7A	S
Myosin, cardiac	MYH7	S
Myotubularin	MTM1	S
Na ⁺ , K ⁺ ATPase, alpha	ATP1A1	G
Na ⁺ , K ⁺ ATPase, beta 1	ATP1B1	G
Na ⁺ , K ⁺ ATPase, beta 2	ATP1B2	G
Na ⁺ , K ⁺ ATPase, beta 3	ATP1B3	G
Na ⁺ /H ⁺ exchanger 1	NHE1	T
Na ⁺ /H ⁺ exchanger 2	NHE2	T
Na ⁺ /H ⁺ exchanger 3	NHE3	T
Na ⁺ /H ⁺ exchanger 4	NHE4	T
Na ⁺ /H ⁺ exchanger 5	NHE5	T
N-acetylgalactosamine-6-sulfate sulfatase	GALNS	E
N-acetylglucosamine-6-sulfatase	GNS	E
N-acetylglucosaminidase, alpha	NAGLU	E
N-acetyltransferase 1	NAT1	E
N-acetyltransferase 2	NAT2	E
NADH dehydrogenase		E

NADH dehydrogenase (ubiquinone) Fe-S protein 1	NDUFS1	E
NADH dehydrogenase (ubiquinone) Fe-S protein 4	NDUFS4	E
NADH dehydrogenase (ubiquinone) flavoprotein 1	NDUFV1	E
NADH-cytochrome b5 reductase	DIA1	E
NADPH-dependent cytochrome P450 reductase	POR	E
Natural resistance-associated macrophage protein 1	NRAMP1	I
NB6		I
Necdin	NDN	G
Nephronophthisis 1	NPHP1	T
Nephronophthisis 2	NPHP2	T
Nephrosis 1	NPHS1	T
Nerve growth factor	NGF	G
Nerve growth factor receptor	NGFR	G
Neural retina-specific gene	NRL	G
Neuraminidase sialidase	NEU	T
Neuregulin	HGL	G
Neurite growth-promoting factor 2	MDK	N
Neurite inhibitory protein		N
Neuroendocrine convertase 1	NEC1, PCSK1	E
Neurofibromin 1	NF1	G
Neurofibromin 2	NF2	G
Neurofilament protein, heavy	NFH	S
Neurofilament protein, NF125	NF150	S
Neurofilament protein, NF200	NF200	S
Neurofilament protein, NF68	NF68	S
Neuronal apoptosis inhibitory protein	NAIP	I
Neuronal molecule-1		I
Neuronal molecule-1 receptor		I
Neuropeptide Y	NPY	N
Neuropeptide Y receptor Y1	NPY1R	N
Neuropeptide Y receptor Y2	NPY2R	N
Neurotrophic tyrosine kinase receptor 1	NTRK1	G
Neurotrophin 3	NTF3 or NT3	G
Neurturin	NRTN	G
Neutral endopeptidase		E
Neutrophil cystolic factor 1	NCF1	I
Neutrophil cystolic factor 2	NCF2	I
Niacin receptor		G
Nibrin	NBS1	G
Nitric oxide synthase 1, NOS1	NOS1	E
Nitric oxide synthase 2, NOS2	NOS2	E
Nitric oxide synthase 3, NOS3	NOS3	E
Nodal	NODAL	G

Noggin	NOG	G
Norrie disease protein	NDP	G
Notch 1	NOTCH1	G
Notch 2	NOTCH2	G
Notch 3	NOTCH3	G
Notch ligand - jagged 1	JAG1, AGS	G
Nuclear factor I-kappa-B-like gene	IKBL	I
Nuclear factor kappa beta	NFKB	I
Nuclear factor of activated T cells (NFAT) complex, cytosolic	NFATC	G
Nuclear factor of activated T cells (NFAT) complex, preexisting component	NFATP	G
Nuclear mitotic apparatus protein 1	NUMA1	G
Nucleophosmin	NPM1	T
Nucleoside diphosphate kinase-A	NDPKA	E
Ocular albinism 1	OA1	S
Oculocutaneous albinism II	OCA2	S
Oligophrenin-1	OPHN1	G
Oncogene abl1	ABL1	G
Oncogene abl2		G
Oncogene akt1		G
Oncogene akt2	AKT2	G
Oncogene axl	AXL	G
Oncogene bcl2		G
Oncogene bcr/abl		G
Oncogene B-lym		G
Oncogene B-raf		G
Oncogene clk1		G
Oncogene c-myc		G
Oncogene cot		G
Oncogene crk		G
Oncogene crkl		G
Oncogene ect2		G
Oncogene ELK1	ELK1	G
Oncogene ELK2	ELK2	G
Oncogene ems1		G
Oncogene ERB		G
Oncogene ERB2		G
Oncogene ERBA		G
Oncogene ERBAL2		G
Oncogene ERG (early reponse gene)		G
Oncogene ETS1		G
Oncogene ETS2		G
Oncogene EVI1	EVI1	G
Oncogene fes		G
Oncogene fgr		G
Oncogene fos	FOS	G
Oncogene fps		G

Oncogene GLI1	GLI	G
Oncogene GLI2	GLI2	G
Oncogene GLI3	GLI3	G
Oncogene gro1		G
Oncogene gro2		G
Oncogene Ha-ras	HRAS	G
Oncogene hst		G
Oncogene hst	FGF4	G
Oncogene int1	WNT1	G
Oncogene int2	FGF3	G
Oncogene int3	Notch4	G
Oncogene int4	WNT3	G
Oncogene jun	JUN	G
Oncogene KIT	KIT, PBT	G
Oncogene LCO	LCO	G
Oncogene l-myc		G
Oncogene lpsa		G
Oncogene lyn		G
Oncogene maf		G
Oncogene mas1		G
Oncogene mcf2		G
Oncogene mdm2	MDM2	G
Oncogene mel		G
Oncogene met	MET	G
Oncogene mos		G
Oncogene mpl		G
Oncogene MUM1	MUM1	G
Oncogene myb	MYB	G
Oncogene myc	MYC	G
Oncogene n-myc		G
Oncogene N-ras (neuroblastoma v-ras)	NRAS	G
Oncogene ovc		G
Oncogene pim1		G
Oncogene pti-1sea		G
Oncogene pvt1		G
Oncogene raf	RAF	G
Oncogene ralb		G
Oncogene rel		G
Oncogene ret	RET	G
Oncogene r-myc		G
Oncogene ros		G
Oncogene R-ras		G
Oncogene sis	PDGFB	G
Oncogene ski		G
Oncogene sno		G
Oncogene spi1		G
Oncogene src		G
Oncogene tc21		G

Oncogene TEL	ETV6	G
Oncogene tim		G
Oncogene vavtrk		G
Oncogene v-Ki-ras2	KRAS2	G
Oncogene yes		G
Oncogene yuasa		G
Oncostatin M	OSM	G
Oncostatin M receptor	OSMR	G
Orexin	OX	G
Orexin 1 receptor	OX1R	G
Orexin 2 receptor	OX2R	G
Ornithine delta-aminotransferase	OAT	E
Ornithine transcarbamoylase	OTC, NME1	E
Orthodenticle (Drosophila) homolog 1	OTX1	G
Orthodenticle (Drosophila) homolog 2	OTX2	G
Osteocalcin		S
Osteonectin	ON	G
Osteopontin	OPN	G
Osteoprotegerin	OPG	G
Otoferlin	OTOF	N
Oxytocin	OXT	N
Oxytocin receptor	OXTR	N
p21-activated kinase 3	PAK3	G
Paired box homeotic gene 1	PAX1	G
Paired box homeotic gene 2	PAX2	G
Paired box homeotic gene 3	PAX3	G
Paired box homeotic gene 6	PAX6	G
Paired box homeotic gene 7	PAX7	G
Paired box homeotic gene 8	PAX8	G
Paired-like homeodomain transcription factor 2	PITX2	G
Paired-like homeodomain transcription factor 3	PITX3	G
Palmitoyl-protein thioesterase	PPT	T
Pancreatic amylase		E
Parathyroid hormone	PTH	G
Parathyroid hormone receptor	PTHr1	G
Parathyroid hormone related-peptide	PTHrP	G
Parathyroid hormone-like hormone	PTHrLH	G
Parvalbumin	PVALB	G
Patched (Drosophila) homolog, PTCH	PTCH	G
PCNA (proliferating cell nuclear antigen)		E
Peanut-like 1	PNUTL1	I
Pendrin, PDS	PDS	T
Peptidylglycine alpha-amidating monooxygenase	PAM	E
Peripherin, PRPH		S
Peroxisomal membrane protein 1	PXMP1	S
Peroxisomal membrane protein 3	PXMP3	T
Peroxisome biogenesis factor 1	PEX1	T

Peroxisome biogenesis factor 19	PEX19	T
Peroxisome biogenesis factor 6	PEX6	T
Peroxisome biogenesis factor 7	PEX7	T
Peroxisome proliferative activated receptor, alpha	PPARA	T
Peroxisome proliferative activated receptor, gamma	PPARG	T
Peroxisome receptor 1	PXR1	T
Phenylethanolamine N-methyltransferase, PNMT	PNMT	E
Phosphatase & tensin homolog	PTEN	G
Phosphate regulating gene with homologies to endopeptidases on the X chromosome	PHEX	G
Phosphatidylinositol glycan, class A (paroxysmal nocturnal hemoglobinuria)	PIGA	G
Phosphatidylinositol transfer protein	PITPN	G
Phosphodiesterase 1 / nucleotide pyrophosphatase 1	PDNP1	G
Phosphodiesterase 1 / nucleotide pyrophosphatase 2	PDNP2	G
Phosphodiesterase 1 / nucleotide pyrophosphatase 3	PDNP3	G
Phosphofructokinase, liver	PFKL	E
Phosphofructokinase, muscle	PFKM	E
Phosphoglucose isomerase	GPI	E
Phosphoglycerate kinase 1	PGK1	E
Phosphoglycerate mutase 2	PGAM2	E
Phospholipase A2, group 10	PLA2G10	I
Phospholipase A2, group 1B	PLA2G1B	I
Phospholipase A2, group 2A	PLA2G2A	I
Phospholipase A2, group 2B	PLA2G2B	I
Phospholipase A2, group 4A	PLA2G4A	I
Phospholipase A2, group 4C	PLA2G4C	I
Phospholipase A2, group 5	PLA2G5	I
Phospholipase A2, group 6	PLA2G6	I
Phospholipase C alpha		I
Phospholipase C beta		I
Phospholipase C delta	PLCD1	I
Phospholipase C epsilon		I
Phospholipase C gamma	PLCG1	I
Phosphomannomutase 1	PMM1	G
Phosphomannomutase 2	PMM2	G
Phosphomannomutase-2	PMM2	T
Phosphorylase kinase deficiency, liver	PHK	E
Phosphorylase kinase, alpha 2	PHKA2	E
Phytanoyl-CoA hydroxylase	PHYH	G
Plakophilin 1	PKP1	T
Plasminogen	PLG	E

Plasminogen activator inhibitor 1	PAI1	E
Plasminogen activator inhibitor 2	PAI2	E
Plasminogen activator receptor, Urokinase	UPAR; PLAUR	S
Plasminogen activator, Tissue	PLAT; TPA	E
Plasminogen activator, Urokinase	UPA; PLAU	E
Platelet derived growth factor	PDGF	G
Platelet derived growth factor receptor	PDGFR	G
Plectin 1	PLEC1	T
Poly (ADP-ribose) synthetase	PARS	E
Poly(A) binding protein 2	PABP2	G
Postsynaptic density-95 protein	PSD95	N
Potassium inwardly-rectifying channel J1	KCNJ1	N
Potassium inwardly-rectifying channel J11	KCNJ11	N
Potassium voltage-gated channel A1	KCNA1	N
Potassium voltage-gated channel E1	KCNE1	N
Potassium voltage-gated channel Q1	KCNQ1	N
Potassium voltage-gated channel Q2	KCNQ2	N
Potassium voltage-gated channel Q3	KCNQ3	N
Potassium voltage-gated channel Q4	KCNQ4	N
POU domain, class 1, transcription factor 1 (Pit1)	POU1F1	G
POU domain, class 3, transcription factor 4	POU3F4	G
POU domain, class 4, transcription factor 3	POU4F3	G
Pre-B-cell leukemia transcription factor 1	PBX1	G
Preproglucagon	GCG;GLP1; GLP2	G
Procollagen N-protease		E
Procollagen peptidase		E
Profibrinolysin		G
Progesterone receptor (RU486 binding receptor)	PGR	G
Prohibitin	PHB	G
Prolactin	PRL	G
Prolactin receptor	PRLR	G
Prolactin releasing hormone	PRH	G
Proliferin	PLF	G
Proline dehydrogenase	PRODH	E
Pro-melanin-concentrating hormone	PMCH	G
Promyelocytic leukemia gene	PML	G
Proopiomelanocortin	POMC	N
Prophet of Pit1	PROP1	G
Propionyl-CoA carboxylase, alpha	PCCA	E
Propionyl-CoA carboxylase, beta	PCCB	E
Prosaposin	PSAP	N
Prostaglandin (PG) D synthase, hematopoietic	PGDS	E
Prostaglandin isomerase		G
Prostaglandin-endoperoxidase synthase 2	PTGS2	G
Prostate cancer anti-metastasis gene KAI1	KAI1	G
Protease nexin 2	PN2	E

Protective protein for beta-galactosidase	PPGB	E
Protein C	PROC	I
Protein kinase A		E
Protein kinase B	PRKB	
Protein kinase C, alpha	PRKCA	E
Protein kinase C, gamma	PRKCG	E
Protein kinase DNA-activated	PRKDC	E
Protein kinase G		E
Protein phosphatase 1, regulatory (inhibitor) subunit 3	PPP1R3	E
Protein phosphatase 2, regulatory subunit A, beta isoform	PPP2R1B	E
Protein tyrosine phosphatase, non-receptor type 12	PTPN12	G
Protoporphyrinogen oxidase	PPOX	E
Pterin-4-alpha-carbinolamine	PCBD	
Purine nucleoside phosphorylase	NP	E
Purinergic receptor P1A1		N
Purinergic receptor P1A2		N
Purinergic receptor P1A3		N
Purinergic receptor P2X, 1	P2RX1	N
Purinergic receptor P2X, 2	P2RX2	N
Purinergic receptor P2X, 3	P2RX3	N
Purinergic receptor P2X, 4	P2RX4	N
Purinergic receptor P2X, 5	P2RX5	N
Purinergic receptor P2X, 6	P2RX6	N
Purinergic receptor P2X, 7	P2RX7	N
Purinergic receptor P2Y, 1	P2RY1	N
Purinergic receptor P2Y, 11	P2RY11	N
Purinergic receptor P2Y, 2	P2RY2	N
Pyrroline-5-carboxylate synthetase	PYCS	E
Pyruvate carboxylase	PC	E
Pyruvate decarboxylase	PDHA	E
Pyruvate kinase	PKLR	E
RAD51, DNA repair protein	RAD51	G
RAD52, DNA repair protein	RAD52	G
RAD54, DNA repair protein	RAD54	G
RAD55, DNA repair protein	RAD55	G
RAD57, DNA repair protein	RAD57	G
Ras-G-protein	RAS	G
Rathke pouch homeobox, RPX	RPX	G
Receptor tyrosine kinase (RTK), Nsk2	NSK2	G
Recombination activating gene 1	RAG1	G
Recombination activating gene 2	RAG2	G
Red cone pigment	RCP	S
Relaxin H1	RLN1	G
Relaxin H2	RLN2	G
Replication factor A		E

Replication factor C	RFC2	E
Retinal pigment epithelium specific protein (65kD)	RPE65	S
Retinitis pigmentosa gene 1	RP1	S
Retinitis pigmentosa gene 2	RP2	S
Retinitis pigmentosa gene 3	RP3	S
Retinitis pigmentosa gene 6	RP6	S
Retinitis pigmentosa gene 7	RP7, RDS	S
Retinoblastoma 1	RB1	G
Retinoic acid receptor, alpha	RARA	G
Retinoic acid receptor, beta	RARB	G
Retinoic acid receptor, gamma	RARG	G
Retinoid X receptor, alpha	RXRA	G
Retinoid X receptor, beta	RXRB	G
Retinoid X receptor, gamma	RXRG	G
Retinoschisis, X-linked, juvenile	RS	G
Rhabdoid tumors	SMARCB1	G
Rhodopsin	RHO	S
Ribonucleotide reductase, RRM		E
Ribosomal protein L13A	RPL13A	G
Ribosomal protein L17	RPL17	G
Ribosomal protein S19	RPS19	E
Ribosomal protein S4, X-linked	RPS4X	E
Ribosomal protein S6 kinase	RPS6KA3	E
Ribosomal protein S9	RPS9	G
RIGUI	RIGUI	G
Rod outer segment membrane protein 1	ROM1	S
Ryanodine receptor 1, skeletal	RYR1	G
SA homolog	SAH	G
Sal-like 1	SALL1	G
Secretin	SCT	T
Semaphorin A4	SEMA4	S
Semaphorin A5	SEMA5	S
Semaphorin D		S
Semaphorin E	SEMAE	S
Semaphorin F	SEMA3/F	S
Semaphorin W	SEMAW	S
Serine/threonine kinase 11	STK11	G
Serine/threonine kinase 2	STK2	G
Serotonin N-acetyltransferase	SNAT	E
Serotonin receptor, 5HT1A	HTR1A	N
Serotonin receptor, 5HT1B	HTR1B	N
Serotonin receptor, 5HT1C	HTR1C	N
Serotonin receptor, 5HT1D	HTR1D	N
Serotonin receptor, 5HT1E	HTR1E	N
Serotonin receptor, 5HT1F	HTR1F	N
Serotonin receptor, 5HT2A	HTR2A	N
Serotonin receptor, 5HT2B	HTR2B	N

Serotonin receptor, 5HT2C	HTR2C	N
Serotonin receptor, 5HT3	HTR3	N
Serotonin receptor, 5HT4	HTR4	N
Serotonin receptor, 5HT5	HTR5	N
Serotonin receptor, 5HT6	HTR6	N
Serotonin receptor, 5HT7	HTR7	N
Serum amyloid A	SAA	T
Serum amyloid P	SAP	T
Sex determining region Y, SRY	SRY	G
Short stature homeobox	SHOX	G
Sialoprotein, bone	BSP	G
Signal transducer and activator of transcription 1	STAT1	G
Signal transducer and activator of transcription 2	STAT2	G
Signal transducer and activator of transcription 3	STAT3	G
Signal transducer and activator of transcription 4	STAT4	G
Signal transducer and activator of transcription 5	STAT5	G
Signaling lymphocyte activation molecule	SLAM	I
Sine oculis homeobox, drosophila, homolog 1	SIX1	G
Sine oculis homeobox, drosophila, homolog 2	SIX2	G
Sine oculis homeobox, drosophila, homolog 5	SIX5	G
Sjogren (Sjogren) syndrome antigen A1	SSA1	I
Slug protein		G
Small nuclear ribonucleoprotein polypeptide N	SNRPN	S
Smoothelin	SMTN	G
Smoothened (Drosophila) homolog	SMOH	G
Sodium channel, non-voltage gated 1, alpha	SCNN1A	N
Sodium channel, non-voltage gated 1, beta	SCNN1B	N
Sodium channel, non-voltage gated 1, gamma	SCNN1G	N
Sodium channel, voltage gated, type IV, alpha polypeptide	SCN4A	N
Sodium channel, voltage gated, type V, alpha polypeptide	SCN5A	N
Sodium channel, voltage-gated, type 1, beta polypeptide	SCN1B	N
Solute carrier family 1 (amino acid transporter), member 6	SLC1A6	T
Solute carrier family 1 (glial high affinity glutamate transporter), member 3	SLC1A3	T
Solute carrier family 1 (glutamate transporter), member 1	SLC1A1	T
Solute carrier family 1 (glutamate transporter), member 2	SLC1A2	T
Solute carrier family 1 (neutral amino acid	SLC1A4	T

transporter), member 4		
Solute carrier family 10 (sodium/bile acid cotransporter family), member 1	SLC10A1	T
Solute carrier family 10 (sodium/bile acid cotransporter family), member 2	SLC10A2	T
Solute carrier family 12, member 1	SLC12A1	T
Solute carrier family 12, member 2	SLC12A2	T
Solute carrier family 12, member 3	SLC12A3	T
Solute carrier family 14, member 2	SLC14A2	T
Solute carrier family 15 (H ⁺ /peptide transporter, intestinal), member 1	SLC15A1	T
Solute carrier family 15 (H ⁺ /peptide transporter, kidney), member 2	SLC15A2	T
Solute carrier family 16 (monocarboxylate transporter), member 1	SLC16A1	T
Solute carrier family 16 (monocarboxylate transporter), member 7	SLC16A7	T
Solute carrier family 17, member 1	SLC17A1	T
Solute carrier family 17, member 2	SLC17A2	T
Solute carrier family 18, member 3	SLC18A3	T
Solute carrier family 19 (folate transporter), member 1	SLC19A1	T
Solute carrier family 2 (facilitated glucose transporter), member 1	SLC2A1	T
Solute carrier family 2 (facilitated glucose transporter), member 2	SLC2A2	T
Solute carrier family 2 (facilitated glucose transporter), member 3	SLC2A3	T
Solute carrier family 2 (facilitated glucose transporter), member 4	SLC2A4	T
Solute carrier family 2 (facilitated glucose transporter), member 5	SLC2A5	T
Solute carrier family 20, member 1	SLC20A1	T
Solute carrier family 20, member 2	SLC20A2	T
Solute carrier family 20, member 3	SLC20A3	T
Solute carrier family 21, member 2	SLC21A2	T
Solute carrier family 21, member 3	SLC21A3	T
Solute carrier family 22, member 1	SLC22A1	T
Solute carrier family 22, member 2	SLC22A2	T
Solute carrier family 22, member 5	SLC22A5	T
Solute carrier family 25, member 12	SLC25A12	T
Solute carrier family 3 (facilitated glucose transporter), member 1	SLC3A1	T
Solute carrier family 4 (anion exchanger), member 1	SLC4A1	T
Solute carrier family 4 (anion exchanger), member 2	SLC4A2	T
Solute carrier family 4 (anion exchanger),	SLC4A3	T

member 3		
Solute carrier family 5 (sodium/glucose transporter), member 1	SLC5A1	T
Solute carrier family 5 (sodium/glucose transporter), member 2	SLC5A2	T
Solute carrier family 5 (sodium/glucose transporter), member 5	SLC5A5	T
Solute carrier family 5, member 3	SLC5A3	T
Solute carrier family 6 (GAMMA-AMINOBUTYRIC ACID transporter), member 1	SLC6A1	T
Solute carrier family 6 (neurotransmitter transporter, dopamine), member 3	SLC6A3	T
Solute carrier family 6 (neurotransmitter transporter, noradrenaline), member 2	SLC6A2	T
Solute carrier family 6 (neurotransmitter transporter, serotonin), member 4	SLC6A4	T
Solute carrier family 6, member 10	SLC6A10	T
Solute carrier family 6, member 6	SLC6A6	T
Solute carrier family 6, member 8	SLC6A8	T
Solute carrier family 7(amino acid transporter), member 1	SLC7A1	T
Solute carrier family 7(amino acid transporter), member 2	SLC7A2	T
Solute carrier family 7(amino acid transporter), member 7	SLC7A7	T
Solute carrier family 8 (sodium/calcium exchanger), member 1	SLC8A1	T
Somatostatin receptor, SSTR2	SSTR2	G
Somatotrophin		G
Sonic hedgehog, SHH	SHH	G
Sorbitol dehydrogenase	SORD	E
Sorcin	SRI	T
SOS1 guanine nucleotide exchange factor	SOS1	G
Spastic paraplegia 7	SPG7	G
Spectrin alpha	SPTA1	S
Spectrin beta	SPTB	S
Sperm adhesion molecule	SPAM1	G
Sperm protamine P1	PRM1	G
Sperm protamine P2	PRM2	G
Sphingomyelinase	SMPD1	E
Spinocerebellar ataxia 8 gene	SCA8	N
Split hand/foot malformation gene	DSS1	G
SRY-box 10	SOX10	G
SRY-box 11	SOX11	G
SRY-box 3	SOX3	G
SRY-box 4	SOX4	G
SRY-box 9	SOX9	G
Stem cell factor	SCF	G

Steroid 5 alpha reductase 1	SRD5A1	E
Steroid 5 alpha reductase 2	SRD5A2	E
Steroid hormone receptor responsive DNA elements		G
Steroid sulphotase	STS	E
Steroidogenic acute regulatory protein	STAR	T
Stromal derived factor 1	SDF1	G
Succinate dehydrogenase 1	SDH1	E
Succinate dehydrogenase 2	SDH2	E
Succinate thiokinase		E
Succinic semi-aldehyde dehydrogenase	ssadh	E
Sulfamidase	SGSH	G
Sulfite oxidase	SUOX	E
Sulfonylurea receptor	SUR	G
Suppression of tumorigenicity 3 gene	ST3	G
Suppression of tumorigenicity 8 gene	ST8	G
Surfactant pulmonary-associated protein A1	SFTPA1	T
Surfactant pulmonary-associated protein A2	SFTPA2	T
Surfactant pulmonary-associated protein B	SFTPB	T
Surfactant pulmonary-associated protein C	SFTPC	T
Surfactant pulmonary-associated protein D	SFTPD	T
Surfeit 1	SURF1	G
Survival of motor neuron 1, telomeric	SMN1	T
SYK-related tyrosine kinase	SRK	I
Syndecan 1	SYND1	G
Syndecan 2	SYND2	G
Syndecan 3	SYND3	G
Syndecan 4	SYND4	G
Synovial sarcoma gene 1	SSX1	G
Synovial sarcoma gene 2	SSX2	G
Talin	TLN	G
TATA binding protein	TBP	G
TATA binding protein associated factor 2A	TAF2A	G
TATA binding protein associated factor 2C2	TAF2C2	G
TATA binding protein associated factor 2D	TAF2E	G
TATA binding protein associated factor 2F	TAF2F	G
TATA binding protein associated factor 2H	TAF2H	G
TATA binding protein associated factor 2I	TAF2I	G
TATA binding protein associated factor 2J	TAF2J	G
TATA binding protein associated factor 2K	TAF2K	G
Tau protein	MAPT	S
T-BOX 1	TBX1	G
T-BOX 2	TBX2	G
T-BOX 3	TBX3	G
T-BOX 4	TBX4	G
T-BOX 5	TBX5	G
T-BOX 6	TBX6	G
T-cell acute lymphocytic leukemia 1	TAL1	I

T-cell acute lymphocytic leukemia 2	TAL2	I
T-cell receptor, alpha	TCRA	I
T-cell receptor, delta	TCRD	I
Telomerase protein component		E
Tenascin (cytotactin)		S
Tenascin XA	TNXA	S
Terminal deoxynucleotidyltransferase, TDT		E
Testis-specific protein Y	TSPY	G
Thiolase, peroxisomal		E
Thiopurine S-methyltransferase	TPMT	E
Thrombopoietin	THPO	G
Thrombospondin	THBS1	G
Thromboxane A synthase 1	TBXAS1	I
Thromboxane A2	TXA2	I
Thromboxane A2 receptor	TBXA2R	I
Thy-1 T-cell antigen	THY1	I
Thymidylate synthase	TYMS	E
Thymopoietin	TMPO	G
Thyroglobulin	TG	G
Thyroid hormone receptor, alpha	THRA	G
Thyroid hormone receptor, beta	THRB	G
Thyroid peroxidase	TPO	G
Thyroid receptor auxiliary protein	TRAP	G
Thyroid-stimulating hormone receptor	TSHR	G
Thyroid-stimulating hormone, alpha	TSHA	G
Thyroid-stimulating hormone, beta	TSHB	G
Thyrotroph embryonic factor	TEF	G
Thyrotropin releasing hormone	TRH	G
Thyrotropin releasing hormone receptor	TRHR	G
Thyroxin-binding globulin	TBG	T
TIE receptor tyrosine kinase	TIE-1	G
Tip-associated protein	TAP	I
Tissue inhibitor of metalloproteinase 1, TIMP1	TIMP1	E
Tissue inhibitor of metalloproteinase 2, TIMP2	TIMP2	E
Tissue inhibitor of metalloproteinase 3, TIMP3	TIMP3	E
Tissue inhibitor of metalloproteinase 4, TIMP4	TIMP4	E
Tissue non-specific alkaline phosphatase		E
TNSAP		
Titin	TTN	S
Tocopherol (alpha) transfer protein	TTPA	T
Toll-like receptor 4	TLR4	I
Topoisomerase I		E
Topoisomerase II		E
Torticollis, keloids, cryptorchidism and renal dysplasia gene	TKCR	G
Transacylase		E
Transcobalamin 1, TCN1		T
Transcobalamin 2, TCN2	TCN2	T

Transcription factor 1, hepatic	TCF1	G
Transcription factor 2, hepatic	TCF2	G
Transcription factor 3	TCF3	G
Transcription factor binding to IGHM enhancer 3	TFE3	G
Transcription factor, TUPLE1	TUPLE1	N
Transcription termination factor, RNA polymerase 1	TTF1	G
Transcription termination factor, RNA polymerase 2	TTF2	G
Transcription termination factor, RNA polymerase 3	TTF3	G
Transferrin	TF	G
Transferrin receptor	TFRC	G
Transforming growth factor, alpha	TGFA	G
Transforming growth factor, beta 2	TGFB2	G
Transforming growth factor, beta induced	TGFB1	G
Transforming growth factor, beta receptor 2	TGFB2R2	G
Transglutaminase 1	TGM1	G
Transglutaminase 2	TGM2	G
Transglutaminase 4	TGM4	G
Transketolase	TKT	E
Transketolase-like 1	TKTL1	E
Translocation in renal carcinoma on chromosome 8 gene	TRC8	G
Transthyretin	TTR	T
Treacle gene	TCOF1	G
Triosephosphate isomerase	TPI1	E
Tropomyosin 1 alpha	TPM1	S
Tropomyosin 3 (non-muscle)	TPM3	S
Troponin C		S
Troponin I	TNNI3	S
Troponin T2, cardiac	TNNT2	S
Trypsin inhibitor		E
Trypsinogen 1	TRY1	E
Trypsinogen 2	TRY2	E
Tryptophan hydroxylase	TPH	E
Tubby-like protein 1	TULP1	G
Tuberous sclerosis 1	TSC1	G
Tuberous sclerosis 2	TSC2	G
Tubulin		S
Tumor susceptibility gene 101	TSG101	G
Tumour necrosis factor (TNF) receptor associated factor 1	TRAF1	I
Tumour necrosis factor (TNF) receptor associated factor 2	TRAF2	I
Tumour necrosis factor (TNF) receptor associated factor 3	TRAF3	I

Tumour necrosis factor (TNF) receptor associated factor 4	TRAF4	I
Tumour necrosis factor (TNF) receptor associated factor 5	TRAF5	I
Tumour necrosis factor (TNF) receptor associated factor 6	TRAF6	I
Tumour necrosis factor alpha	TNFA	I
Tumour necrosis factor alpha receptor	TNFAR	I
Tumour necrosis factor beta	TNFB	I
Tumour necrosis factor beta receptor	TNFBR	I
Tumour protein p53	TP53, P53	G
Tumour protein p63	TP63	G
Tumour protein p73	TP73	G
Tumour protein, translationally-controlled 1	TPT1	G
Tumour suppressor gene DRA	DRA	I
Twist (Drosophila) homolog	TWIST	G
Tyrosinase	TYR	E
Tyrosinase-related protein 1	TYRP1	E
Tyrosine aminotransferase	TAT	E
Tyrosine hydroxylase	TH	E
Ubiquitin		E
Ubiquitin activating enzyme, E1		G
Ubiquitin B	UBB	E
Ubiquitin C	UBC	G
Ubiquitin carboxyl-terminal esterase L1	UCHL1	G
Ubiquitin fusion degeneration 1-like	UFD1L	G
Ubiquitin protein ligase E3A	UBE3A	E
UDP-glucose pyrophosphorylase		E
UDP-glucuronosyltransferase 1	ugt1d, UGT1	E
UDP-glucuronosyltransferase 2	UGT2	E
Uncoupling protein 1		T
Uncoupling protein 3	UCP3	T
Undulin 1	COL14A1	S
Uridine monophosphate kinase	UMPK	I
Uridine monophosphate synthetase	UMPS	I
Uridinediphosphate(UDP)-galactose-4-epimerase	GALE	E
Uroporphyrinogen decarboxylase	UROD	E
Uroporphyrinogen III synthase	UROS	E
Usher syndrome 2A	USH2A	S
Vascular endothelial growth factor	VEGF	G
Vasoinhibitory peptide		G
Vitamin B12-binding (R) protein		G
Vitamin D receptor	VDR	G
Vitelliform macular dystrophy, atypical gene	VMD1	T
v-myc avian myelocytomatosis viral oncogene homolog	MYC	G
Von Hippel-Lindau gene	VHL	G

Werner syndrome helicase	WRN	G
Wilms tumour gene 1	WT1	G
Wilms tumour gene 2	WT2	G
Wilms tumour gene 4	WT4	G
Winged helix nude	WHN	G
Wingless family, wnt2	WNT2	G
Wingless family, wnt4	WNT4	G
Wingless family, wnt5	WNT5	G
Wingless family, wnt7	WNT7	G
Wingless family, wnt8	WNT8	G
Wiskott-Aldrich syndrome protein	WASP, THC	I
Wnt inhibitory factor, WIF-1	WIF1	G
Wolf-Hirschhorn syndrome candidate 1 gene	WHSC1	G
Wolfram syndrome 1 gene	WFS1	S
X (inactive)-specific transcript	XIST	G
Xanthine dehydrogenase	XDH	E
Xeroderma pigmentosum, complementation group A	XPA	E
Xeroderma pigmentosum, complementation group B	XPB	E
Xeroderma pigmentosum, complementation group C	XPC	E
Xeroderma pigmentosum, complementation group D		E
Xeroderma pigmentosum, complementation group E		E
Xeroderma pigmentosum, complementation group F	XPF	E
Xeroderma pigmentosum, complementation group G	ERCC5	E
X-ray repair gene	XRCC9	G
Xylitol dehydrogenase		E
YY1 transcription factor	YY1	G
Zinc finger protein 198	ZIC198	S
Zinc finger protein 2	ZIC2	S
Zinc finger protein 3	ZIC3	S
Zinc finger protein HRX	ALL1	I
Zona pellucida glycoprotein 1	ZP1	G
Zona pellucida glycoprotein 2	ZP2	G
Zona pellucida glycoprotein 3	ZP3	G
Zona pellucida receptor tyrosine kinase	ZRK	G
Zonadhesin	ZAN	G

CLAIMS

1. A set of nucleotide probes for detecting relevant variants (mutations and polymorphisms), e.g. nucleotide substitutions (missense, nonsense, splicing and regulatory), small deletions, small insertions, small insertion deletions, gross insertions, gross deletions, duplications, complex rearrangements and repeat variations in a target group of genes which relate to adverse events; said probes being complementary to DNA and RNA sequences of said group of genes; characterised in that said group is a core group of genes consisting of substantially all of the following:

KEY TO 'PROTEIN FUNCTION' COLUMN

E ENZYME
 T TRANSPORT & STORAGE
 S STRUCTURAL
 I IMMUNITY
 N NERVOUS TRANSMISSION
 G GROWTH & DIFFERENTIATION

ADME GENE LIST

	HUGO gene symbol	Protein function
5-adenosyl homocysteine hydrolase		E
Acetoacetyl 1-CoA-thiolase	ACAT1	E
Acetoacetyl 2-CoA-thiolase	ACAT2	E
Acetyl CoA acyltransferase	ACAA	E
Acetylcholine receptor, nicotinic, alpha A1	CHRNA1	N
Acetylcholine receptor, nicotinic, alpha A2	CHRNA2	N
Acetylcholine receptor, nicotinic, alpha A3	CHRNA3	N
Acetylcholine receptor, nicotinic, alpha A4	CHRNA4	N
Acetylcholine receptor, nicotinic, alpha A5	CHRNA5	N
Acetylcholine receptor, nicotinic, alpha A6	CHRNA6	N
Acetylcholine receptor, nicotinic, alpha A7	CHRNA7	N
Acetylcholine receptor, nicotinic, beta 1	CHRNA1	N
Acetylcholine receptor, nicotinic, beta 2	CHRNA2	N
Acetylcholine receptor, nicotinic, beta 3	CHRNA3	N
Acetylcholine receptor, nicotinic, beta 4	CHRNA4	N
Acetylcholine receptor, nicotinic, epsilon	CHRNA5	N
Acetylcholine receptor, nicotinic, gamma	CHRNA6	N
Acetylcholinesterase	ACHE	E
Actin, alpha, cardiac	ACTC	S
Actin, alpha, skeletal	ACTA1	S
Actin, alpha, smooth, aortic	ACTA2	S
Actin, beta	ACTB	S
Actin, gamma 2	ACTG2	S
Acyl CoA dehydrogenase, short chain	ACADS	E

Adenine phosphoribosyltransferase	APRT	T
Adenosine deaminase	ADA	E
Adenosine monophosphate deaminase	AMPD	E
Adenosine receptor A1	ADORA1	N
Adenosine receptor A2A	ADORA2A	N
Adenosine receptor A2B	ADORA2B	N
Adenosine receptor A3	ADORA3	N
Adenylate cyclase 1	ADCY1	E
Adenylate cyclase 2	ADCY2	E
Adenylate cyclase 3	ADCY3	E
Adenylate cyclase 4	ADCY4	E
Adenylate cyclase 5	ADCY5	E
Adenylate cyclase 6	ADCY6	E
Adenylate cyclase 7	ADCY7	E
Adenylate cyclase 8	ADCY8	E
Adenylate cyclase 9	ADCY9	E
Adenylate kinase	AK1	E
Adenylate transferase		E
Adenylosuccinate lyase	ADSL	E
ADP-ribosyltransferase	ADPRT	E
Adrenergic receptor, alpha1	ADRA1	N
Adrenergic receptor, alpha2	ADRA2	N
Adrenergic receptor, beta1	ADRB1	N
Adrenergic receptor, beta2	ADRB2	N
Adrenergic receptor, beta3	ADRB3	N
Adrenocorticotrophic hormone (ACTH) receptor	ACTHR	G
Adrenoleukodystrophy gene	ALD	E
Albumin, ALB	ALB	T
Alkaptonuria gene	AKU	G
Alpha 1 acid glycoprotein	AAG; AGP	T
alpha1-antitrypsin	PI	E
alpha2-antiplasmin	PLI	E
alpha-amylase		E
Alpha-fetoprotein	AFP	G
alpha-glucosidase, neutral AB	GANAB	E
alpha-glucosidase, neutral C	GANC	E
Aminomethyltransferase	AMT	E
Aminopeptidase P	XPNPEP2	E
Amyloid beta (A4) precursor protein-binding, APBB1	APBB1	N
Amyloid beta A4 precursor protein	APP	N
Androgen binding protein	ABP	T
Androgen receptor	AR	G
Angiotensin converting enzyme	ACE, DCP1	E
Angiotensin receptor 1	AGTR1	T
Angiotensin receptor 2	AGTR2	T
Angiotensinogen	AGT	E

Annexin 1	ANX 1	I
Apurinic endonuclease	APE	E
Arginine vasopressin	AVP	N
Arginine vasopressin receptor 1A	AVPR1A	N
Arginine vasopressin receptor 1B	AVPR1B	N
Arginine vasopressin receptor 2	AVPR2	N
Aryl hydrocarbon receptor	AHR	T
Arylsulfatase E	ARSE	E
Aspartate transcarbamoylase		E
Ataxia telangiectasia gene, AT	ATM	G
ATP cobalamin adenoxyltransferase		E
ATP sulphurylase	atpsk2	E
ATP/ADP translocase		E
Atrial natriuretic peptide	ANP	G
Atrial natriuretic peptide receptor A	NPR1	G
Atrial natriuretic peptide receptor B	NPR2	G
Atrial natriuretic peptide receptor C	NPR3	G
BCL2-associated X protein	BAX	G
Benzodiazepine receptor		N
beta-endorphin receptor		N
Bile acid coenzyme A: amino acid N-acyltransferase	BAAT	E
Bile salt export pump	BSEP, PFIC2	T
Bile salt-stimulated lipase	CEL	E
Bilirubin UDP-glucuronosyltransferase		E
Biliverdin reductase		T
Bleomycin hydrolase	BLMH	E
Bradykinin receptor B1		I
Bradykinin receptor B2		I
Breakpoint cluster region	BCR	G
Breast cancer 1	BRCA1	G
Breast cancer 2	BRCA2	G
Brush border guanylyl cyclase		E
Butyrylcholinesterase	BCHE	E
Ca(2+) transporting ATPase, fast twitch	ATP2A1	T
Ca(2+) transporting ATPase, slow twitch	ATP2A2	T
Calcineurin A1	CALNA1	I
Calcineurin A2	CALNA2	I
Calcineurin A3	CALNA3	I
Calcineurin B		I
Calcitonin receptor /Calcitonin gene-related peptide receptor	CALCR	N
Calcium channel, voltage-dependent, alpha 1F subunit	CACNA1F	N
Calcium channel, voltage-dependent, Alpha-1B (CACNL1A5)	CACNA1B	N
Calcium channel, voltage-dependent, Alpha-1C	CACNA1C	N

Calcium channel, voltage-dependent, Alpha-1D	CACNA1D	N
Calcium channel, voltage-dependent, Alpha-1E (CACNL1A6)	CACNA1E	N
Calcium channel, voltage-dependent, Alpha-2/delta	CACNA2	N
Calcium channel, voltage-dependent, Beta 1	CACNB1	N
Calcium channel, voltage-dependent, Beta 3	CACNB3	N
Calcium channel, voltage-dependent, L type, alpha 1S subunit	CACNA1S	N
Calcium channel, voltage-dependent, Neuronal, Gamma	CACNG2	N
Calcium channel, voltage-dependent, P/Q type, alpha 1A subunit	CACNA1A	N
Calcium channel, voltage-dependent, T-type		N
Canalicular multispecific organic anion transporter	CMOAT	T
Cannabinoid receptor	CNR1	N
Carbamoylphosphate synthetase 1	CPS1	E
Carbamoylphosphate synthetase 2	CPS2	E
Carbonic anhydrase 3	CA3	E
Carbonic anhydrase 4	CA4	E
Carbonic anhydrase, alpha	CA1	E
Carbonic anhydrase, beta	CA2	E
Carnitine transporter protein	CDSP, SCD	T
Carnosinase		N
Cartilage-hair hypoplasia gene	CHH	N
Catalase	CAT	I
Catechol-O-methyltransferase	COMT	E
Catenin, beta	CTNNB1	G
Cell adhesion molecule, vascular, VCAM	VCAM1	G
Cholecystokinin	CCK	N
Cholecystokinin B receptor	CCKBR	N
Cholesterol ester transfer protein	CETP	T
Choline acetyltransferase	CHAT	E
CoA transferase		E
Colony-stimulating factor 1	CSF1	G
Colony-stimulating factor 2	CSF2	G
Colony-stimulating factor 3	CSF3	G
Colony-stimulating factor 3 receptor	CSF3R	G
Complex V	MTATP6	E
Coproporphyrinogen oxidase	CPO	E
Cortico-steroid binding protein		T
Corticosteroid nuclear receptor		I
Corticotrophin-releasing hormone receptor	CRHR1	T
Creb binding protein	CREBBP	G
Crystallin, alpha A	CRYAA	S
Crystallin, alpha B	CRYAB	S

Crystallin, beta B2	CRYBB2	S
Crystallin, gamma A	CRYGA	S
Cu ²⁺ transporting ATPase alpha polypeptide	ATP7A	E
Cu ²⁺ transporting ATPase beta polypeptide	ATP7B	E
Cyclic AMP response element binding protein	CREB	G
Cyclic AMP response element modulator	CREM	G
Cyclic AMP-dependent protein kinase	PKA	E
Cyclic nucleotide phosphodiesterase 1B	PDE1B	E
Cyclic nucleotide phosphodiesterase 1B1	PDE1B1	E
Cyclic nucleotide phosphodiesterase 2A3	PDE2A3	E
Cyclic nucleotide phosphodiesterase 3A	PDE3A	E
Cyclic nucleotide phosphodiesterase 3B	PDE3B	E
Cyclic nucleotide phosphodiesterase 4A	PDE4A	E
Cyclic nucleotide phosphodiesterase 4C	PDE4C	E
Cyclic nucleotide phosphodiesterase 5A	PDE5A	E
Cyclic nucleotide phosphodiesterase 6A	PDE6A	E
Cyclic nucleotide phosphodiesterase 6B	PDE6B	E
Cyclic nucleotide phosphodiesterase 7	PDE7	E
Cyclic nucleotide phosphodiesterase 8	PDE8	E
Cyclic nucleotide phosphodiesterase 9A	PDE9A	E
Cyclin F	CCNF	G
Cyclin-dependent kinase inhibitor 1A (P21, CIP1)	CDKN1A	G
Cyclooxygenase 1	COX1	E
Cyclooxygenase 2	COX2	E
Cyclophilin		I
CYP11A1	CYP11A1	E
CYP11B1	CYP11B1	E
CYP11B2	CYP11B2	E
CYP17	CYP17	E
CYP19	CYP19	E
CYP1A1	CYP1A1	E
CYP1A2	CYP1A2	E
CYP1B1	CYP1B1	E
CYP21	CYP21	E
CYP24	CYP24	E
CYP27	CYP27	E
CYP27B1	PDDR	E
CYP2A1	CYP2A1	E
CYP2A13	CYP2A13	E
CYP2A3	CYP2A3	E
CYP2A6V2	CYP2A6V2	E
CYP2A7	CYP2A7	E
CYP2B6	CYP2B6	E
CYP2C18	CYP2C18	E
CYP2C19	CYP2C19	E
CYP2C8	CYP2C8	E
CYP2C9	CYP2C9	E

CYP2D6	CYP2D6	E
CYP2E1	CYP2E1	E
CYP2F1	CYP2F1	E
CYP2J2	CYP2J2	E
CYP3A3	CYP3A3	E
CYP3A4	CYP3A4	E
CYP3A5	CYP3A5	E
CYP3A7	CYP3A7	E
CYP4A11	CYP4A11	E
CYP4B1	CYP4B1	E
CYP4F2	CYP4F2	E
CYP4F3	CYP4F3	E
CYP51	CYP51	E
CYP5A1	CYP5A1	E
CYP7A	CYP7A	E
CYP8	CYP8	E
Cystic fibrosis transmembrane conductance regulator, CFTR	CFTR	N
Cytidine deaminase	CDA	E
Cytidine-5-prime-triphosphate synthetase	CTPS	E
Cytokine-suppressive antiinflammatory drug- binding protein 1	CSBP1	I
Cytokine-suppressive antiinflammatory drug- binding protein 2	CSBP2	I
Deoxycytidine kinase DCK		E
Deoxyuridine triphosphatase; dUTPase		E
DHEA sulfotransferase	STD	E
Dihydrodiol dehydrogenase 1	DDH1	E
Dihydrofolate reductase	DHFR	E
Dihydrolipoamide branched chain transacylase	DBT	N
Dihydrolipoamide dehydrogenase	DLD	N
Dihydrolipoyl dehydrogenase 2	PDHA	E
Dihydrolipoyl transacetylase	PDHA	E
Dihydroorotase		E
Dihydropyrimidine dehydrogenase	DPYD	E
Disrupted meiotic cDNA 1, homolog	DMC1	G
DNA damage binding protein, DDB1	DDB1	S
DNA damage binding protein, DDB2	DDB2	S
DNA directed polymerase, alpha	POLA	E
DNA glycosylases		E
DNA helicases		E
DNA Ligase 1	LIG1	E
DNA methyltransferase	DNMT	E
DNA polymerase 1		E
DNA polymerase 2		E
DNA polymerase 3		E
DNA primase		E
DNA-damage-inducible transcript 3	DDIT3	S

DNA-dependant RNA polymerase		E
Dopamine receptors D1	DRD1	N
Dopamine receptors D2	DRD2	N
Dopamine receptors D3	DRD3	N
Dopamine receptors D4	DRD4	N
Dopamine receptors D5	DRD5	N
Erythropoietin	EPO	I
Erythropoietin receptor	EPOR	I
Estrogen receptor	ESR	G
Excision repair complementation group 1 protein	ERCC1	E
Excision repair complementation group 2 protein	ERCC2	E
Excision repair complementation group 2 protein	ERCC3	E
Excision repair complementation group 4 protein	ERCC4	E
Excision repair complementation group 6 protein	ERCC6	E
Factor H	HF1	I
Factor IX	F9	I
Factor VII	F7	I
Factor VIII	F8	I
Factor X	F10	I
Fatty acid binding proteins FABP1		T
Fatty acid binding proteins FABP2	FABP2	T
Fatty acid binding proteins FABP3		T
Fatty acid binding proteins FABP4		T
Fatty acid binding proteins FABP5		T
Fatty acid binding proteins FABP6		T
Fibroblast growth factor	FGF1	G
Flavin-containing monooxygenase 1	FMO1	E
Flavin-containing monooxygenase 2	FMO2	E
Flavin-containing monooxygenase 3	FMO3	E
Flavin-containing monooxygenase 4	FMO4	E
Folic acid receptor	FOLR	G
Follicle stimulating hormone receptor	FSHR, ODG1	G
Follicle stimulating hormone, FSH	FSHB	G
Forkhead transcription factor 10	FKHL10	G
Forkhead transcription factor 14	FKHL14	G
Forkhead transcription factor 7	FKHL7	G
G/T mismatch binding protein	GTBP, MSH6	G
GABA receptor, alpha 1	GABRA1	N
GABA receptor, alpha 2	GABRA2	N
GABA receptor, alpha 3	GABRA3	N
GABA receptor, alpha 4	GABRA4	N
GABA receptor, alpha 5	GABRA5	N
GABA receptor, alpha 6	GABRA6	N

GABA receptor, beta 1	GABRB1	N
GABA receptor, beta 2	GABRB2	N
GABA receptor, beta 3	GABRB3	N
GABA receptor, gamma 1	GABRG1	N
GABA receptor, gamma 2	GABRG2	N
GABA receptor, gamma 3	GABRG3	N
GABA transaminase	ABAT	E
Gadd45 (growth arrest & DNA-damage-inducible protein)		E
Galactose 1-phosphate uridyl-transferase	GALT	E
Gamma-glutamyl carboxylase	GGCX	T
Gamma-glutamyltransferase 1	GGT1	T
Gamma-glutamyltransferase 2	GGT2	T
Gastric inhibitory polypeptide receptor, GIPR	GIPR	T
Gastric lipase, LIPF		T
Glucagon receptor	GCGR	G
Glucocorticoid receptor	GRL	G
Glucosaminyl (N-acetyl) transferase 2, 1-branching enzyme	GCNT2	E
Glucosidase, acid beta	GBA	E
Glutamate decarboxylase, GAD	GAD1	E
Glutamate receptor 1	GLUR1	N
Glutamate receptor 2	GLUR2	N
Glutamate receptor 3	GLUR3	N
Glutamate receptor 4	GLUR4	N
Glutamate receptor 5	GLUR5	N
Glutamate receptor 6	GLUR6	N
Glutamate receptor 7	GLUR7	N
Glutamate receptor, ionotropic, NMDA 1	NMDAR1	N
Glutamate receptor, ionotropic, NMDA 2A	NMDAR2A	N
Glutamate receptor, ionotropic, NMDA 2B	NMDAR2B	N
Glutamate receptor, ionotropic, NMDA 2C	NMDAR2C	N
Glutamate receptor, ionotropic, NMDA 2D	NMDAR2D	N
Glutamine phosphoribosylpyrophosphate amidotransferase/PRPP amidotransferase		E
Glutathione	GSH	T
Glutathione peroxidase, GPX1	GPX1	E
Glutathione peroxidase, GPX2	GPX2	E
Glutathione reductase, GSR	GSR	E
Glutathione S-transferase mu 1, GSTM1	GSTM1	E
Glutathione S-transferase mu 4, GSTM4		E
Glutathione S-transferase theta 1, GSTT1	GSTT1	E
Glutathione S-transferase theta 2, GSTT2		E
Glutathione S-transferase, GSTP1	GSTP1	E
Glutathione S-transferase, GSTZ1	GSTZ1	E
Glutathione synthetase	GSS	E
Glyceraldehyde-3-phosphate dehydrogenase, GAPDH	GAPDH	E
Glycinamide ribonucleotide (GAR)	GART	E

transformylase		
Glycine receptor, alpha	GLRA2	N
Glycine receptor, beta		N
Glycine transporter	GLYT	N
Gonadotropin releasing hormone	GNRH	G
Gonadotropin releasing hormone receptor	GNRHR	G
Growth arrest-specific homeobox	GAX	G
Growth hormone 1	GH1	G
Growth hormone 2 (placental)	GH2	G
Growth hormone receptor	GHR	G
Growth hormone releasing hormone (GHRH)	GHRH	G
Growth hormone releasing hormone receptor	GHRHR	G
GTP cylohydrolase 1	GCH1	G
GTPase-activating protein, GAP	RASA1	G
Guanidinoacetate N-methyltransferase	GAMT	E
Guanine nucleotide-binding protein, alpha activating activity polypeptide, GNAO	GNAO1	N
Guanine nucleotide-binding protein, alpha inhibiting activity polypeptide 1, GNAI1	GNAI1	N
Guanine nucleotide-binding protein, alpha inhibiting activity polypeptide 2, GNAI2	GNAI2	N
Guanine nucleotide-binding protein, alpha inhibiting activity polypeptide 3, GNAI3	GNAI3	N
Guanine nucleotide-binding protein, alpha stimulating activity polypeptide, GNAS1	GNAS1	N
Guanine nucleotide-binding protein, alpha stimulating activity polypeptide, GNAS2	GNAS2	N
Guanine nucleotide-binding protein, alpha stimulating activity polypeptide, GNAS3	GNAS3	N
Guanine nucleotide-binding protein, alpha stimulating activity polypeptide, GNAS4	GNAS4	N
Guanine nucleotide-binding protein, alpha transducing activity polypeptide, GNAT1	GNAT1	N
Guanine nucleotide-binding protein, alpha transducing activity polypeptide, GNAT2	GNAT2	N
Guanine nucleotide-binding protein, beta polypeptide 3	GNB3	N
Guanine nucleotide-binding protein, gamma polypeptide 5	GNG5	N
Guanine nucleotide-binding protein, q polypeptide	GNAQ	N
Guanylate cyclase 2D, membrane (retina-specific)	GUCY2D	E
Guanylate cyclase activator 1A (retina)	GUCA1A	E
Guanylate kinase		E
Guanylin	GUCA2	T
Guanylyl cyclase		E
H(+), K(+) - ATPase	ATP4B	N

Heat shock protein, HSP60		I
Heat shock protein, HSP70		I
Heat shock protein, HSP90		I
Hemopexin	HPX	I
Hepatic lipase	LIPC	I
Histamine receptors, H1		E
Histamine receptors, H2		N
Histamine receptors, H3		N
HLH transcription factor HAND1	HAND1	N
HLH transcription factor HAND2	HAND2	G
HMG-CoA lyase	HMGCL	G
HMG-CoA reductase	HMGCR	E
HMG-CoA synthase	HMGCS2	E
Hormone-sensitive lipase	HSL	E
HSSB, replication protein		E
Hypoxanthine-guanine phosphoribosyltransferase, HGPRT	HPRT	E
Ibonucleoside diphosphate reductase		E
Ikaros gene	IKAROS	E
Inosine monophosphate dehydrogenase, IMPDH		G
Inosine triphosphatase	ITPA	E
Inositol monophosphatase	IMPA1	N
Insulin	INS	G
Insulin receptor	INSR	G
Insulin-like growth factor 1 receptor	IGF1R	G
Insulin-like growth factor 2 receptor	IGF2R	G
Interferon alpha	IFNA1	I
Interferon beta	IFNB	I
Interferon gamma	IFNG	I
Interferon gamma receptor 1	IFNGR1	I
Interferon gamma receptor 2	IFNGR2	I
Interferon regulatory factor 1	IRF1	I
Interferon regulatory factor 4	IRF4	I
Interleukin(IL) 1 receptor	IL1R	I
Interleukin(IL) 1, alpha	IL1A	I
Interleukin(IL) 1, beta	IL1B	I
Interleukin(IL) 10	IL10	I
Interleukin(IL) 10 receptor	IL10R	I
Interleukin(IL) 11	IL11	I
Interleukin(IL) 11 receptor	IL11R	I
Interleukin(IL) 12	IL12	I
Interleukin(IL) 12 receptor, beta 1	IL12RB1	I
Interleukin(IL) 13	IL13	I
Interleukin(IL) 13 receptor	IL13R	I
Interleukin(IL) 2	IL2	I
Interleukin(IL) 2 receptor, alpha	IL2RA	I
Interleukin(IL) 2 receptor, gamma	IL2RG	I

Interleukin(IL) 3	IL3	I
Interleukin(IL) 3 receptor	IL3R	I
Interleukin(IL) 4	IL4	I
Interleukin(IL) 4 receptor	IL4R	I
Interleukin(IL) 5	IL5	I
Interleukin(IL) 5 receptor	IL5R	I
Interleukin(IL) 6	IL6	I
Interleukin(IL) 6 receptor	IL6R	I
Interleukin(IL) 7	IL7	I
Interleukin(IL) 7 receptor	IL7R	I
Interleukin(IL) 8	IL8	I
Interleukin(IL) 8 receptor	IL8R	I
Interleukin(IL) 9	IL9	I
Interleukin(IL) 9 receptor	IL9R	I
Interleukin(IL) receptor antagonist 1	IL1RN, IL1RA	I
Kallikrein 3	KAK3	I
Kinectin	KTN1	G
Kinesin, heavy chain	KNSL1	G
Kinesin, light chain	KNS2	G
Kininogen, High molecular weight	KNG	I
Leptin	LEP	G
Leptin receptor	LEPR	G
Leukotriene A4 hydrolase		I
Leukotriene B4 receptor		I
Leukotriene C4 receptor		I
Leukotriene D4/E4 receptor		I
LH/choriogonadotropin (CG) receptor	LHCGR	G
LIM homeobox transcription factor 1, beta	LMX1B	G
Lipoprotein lipase	LPL	I
Lipoprotein receptor, Low Density	LDLR	T
Lipoxygenase 12 (platelets)	LOG12	I
Lipoxygenase 5 (leukocytes)		I
Low density lipoprotein receptor-related protein LRP precursor		T
Lysosomal acid lipase	LIPA	E
Malonyl CoA decarboxylase		E
Malonyl CoA transferase		E
Maltase-glucoamylase		E
Mannose binding protein	MBP	I
Mannosyl (alpha-1,6-)-glycoprotein beta-1, 2-	MGAT2	T
N-acetylglucosaminyltransferase		
MAPK kinase 1	MAPKK1; MEK1	G
MAPK kinase 4	MAPKK4; MEK4;	G
	SERK1	
MAPK kinase 6	MAPKK6; MEK6	G
MAPKK kinase	MAPKKK	G
Matrix Gla protein	MGP	G
MEK kinase, MEKK		E

Melanocortin 2 receptor	MC2R	T
Melanocortin 4 receptor	MC4R	T
Methionine adenosyltransferase	MAT1A, MAT2A	E
Methionine synthase	MTR	E
Methionine synthase reductase	MTRR	E
Methylguanine-DNA methyltransferase	MGMT	E
Mevalonate kinase	MVK	E
MHC Class I: Tap1	ABCR, TAP1	I
MHC Class II: Tap2	TAP2, PSF2	I
Microphthalmia-associated transcription factor	MITF	G
Mismatch repair gene, PMSL1	PMS1	G
Mismatch repair gene, PMSL2	PMS2	G
Mitochondrial trifunctional protein, alpha subunit	HADHA	E
Mitochondrial trifunctional protein, beta subunit	HADHB	E
Mitogen-activated protein (MAP) kinase	MAPK	G
Monoamine oxidase A	MAOA	E
Monoamine oxidase B	MAOB	E
Multidrug resistance associated protein	MRP	G
Muscarinic receptor, M1	CHRM1	N
Muscarinic receptor, M2	CHRM2	N
Muscarinic receptor, M3	CHRM3	N
Muscarinic receptor, M4	CHRM4	N
Muscarinic receptor, M5	CHRM5	N
Na ⁺ , K ⁺ ATPase, alpha	ATP1A1	G
Na ⁺ , K ⁺ ATPase, beta 1	ATP1B1	G
Na ⁺ , K ⁺ ATPase, beta 2	ATP1B2	G
Na ⁺ , K ⁺ ATPase, beta 3	ATP1B3	G
Na ⁺ /H ⁺ exchanger 1	NHE1	T
Na ⁺ /H ⁺ exchanger 2	NHE2	T
Na ⁺ /H ⁺ exchanger 3	NHE3	T
Na ⁺ /H ⁺ exchanger 4	NHE4	T
Na ⁺ /H ⁺ exchanger 5	NHE5	T
N-acetylgalactosamine-6-sulfate sulfatase	GALNS	E
N-acetylglucosamine-6-sulfatase	GNS	E
N-acetylglucosaminidase, alpha	NAGLU	E
N-acetyltransferase 1	NAT1	E
N-acetyltransferase 2	NAT2	E
N-acyl hydrolase		E
NADH dehydrogenase (ubiquinone) flavoprotein 1	NDUFV1	I
NADH-cytochrome b5 reductase	DIA1	E
NADPH-dependent cytochrome P450 reductase	POR	E
Nephrolithiasis 2	NPHL2	T
Nephronophthisis 2	NPHP2	T
Nephrosis 1	NPHS1	T
Neuroendocrine convertase 1	NEC1, PCSK1	E

Neurokinin A	NKNA	N
Neurokinin B	NKNB	N
Neuropeptide Y	NPY	N
Neuropeptide Y receptor Y1	NPY1R	N
Neuropeptide Y receptor Y2	NPY2R	N
Niacin receptor		G
Niemann-Pick disease protein	NPC1	T
Nuclear factor kappa beta	NFKB	I
Nuclear factor of activated T cells (NFAT) complex, cytosolic	NFATC	G
Nuclear factor of activated T cells (NFAT) complex, preexisting component	NFATP	G
Nucleoside diphosphate kinase-A	NDPKA	E
Oncogene spi1		G
Opioid receptor, delta	OPRD1	N
Opioid receptor, kappa	OPRK1	N
Opioid receptor, mu	OPRM1	N
Ornithine transcarbamoylase	OTC, NME1	E
Osteoprotegerin	OPG	G
Otoferlin	OTOF	N
Oxytocin	OXT	N
Oxytocin receptor	OXTR	N
Paired-like homeodomain transcription factor 2	PITX2	G
Paired-like homeodomain transcription factor 3	PITX3	G
Paraoxonase PON1	PON1	E
Paraoxonase PON2	PON2	E
Paraoxonase PON3		E
Parathyroid hormone	PTH	G
Parathyroid hormone receptor	PTHr1	G
Parathyroid hormone related-peptide	PTHrP	G
Parathyroid hormone-like hormone	PTHLH	G
Parvalbumin	PVALB	G
PCNA (proliferating cell nuclear antigen)		E
Peanut-like 1	PNUTL1	I
Peroxisomal membrane protein 1	PXMP1	S
Peroxisome biogenesis factor 1	PEX1	T
Peroxisome biogenesis factor 19	PEX19	T
Peroxisome biogenesis factor 6	PEX6	T
Peroxisome biogenesis factor 7	PEX7	T
Peroxisome proliferative activated receptor, alpha	PPARA	T
Peroxisome proliferative activated receptor, gamma	PPARG	T
P-glycoprotein 1	PGY1	T
P-glycoprotein 3	PGY3	T
Phenylethanolamine N-methyltransferase, PNMT	PNMT	E
Phosphodiesterase 1 / nucleotide	PDNP1	G

pyrophosphatase 1		
Phosphodiesterase 1 / nucleotide pyrophosphatase 2	PDNP2	G
Phosphodiesterase 1 / nucleotide pyrophosphatase 3	PDNP3	G
Phospholipase A2, group 10	PLA2G10	I
Phospholipase A2, group 1B	PLA2G1B	I
Phospholipase A2, group 2A	PLA2G2A	I
Phospholipase A2, group 2B	PLA2G2B	I
Phospholipase A2, group 4A	PLA2G4A	I
Phospholipase A2, group 4C	PLA2G4C	I
Phospholipase A2, group 5	PLA2G5	I
Phospholipase A2, group 6	PLA2G6	I
Phospholipase C alpha		I
Phospholipase C beta		I
Phospholipase C delta	PLCD1	I
Phospholipase C epsilon		I
Phospholipase C gamma	PLCG1	I
Phosphomannomutase-2	PMM2	T
Phosphomannose isomerase-1, PMI1	MPI	T
Phosphoribosyl pyrophosphate synthetase	PRPS1	E
Pituitary adenylate cyclase activating peptide	PACAP	N
Pituitary adenylate cyclase activating peptide receptor	PACAP1R	N
Plasminogen activator, Tissue	PLAT; TPA	E
Platelet-activating factor receptor	PAFR	I
Plectin 1	PLEC1	T
Polycystin 1	PKD1	T
Polycystin 2	PKD2	T
Porphobilinogen deaminase	HMBS	E
Potassium channel, calcium-activated,	KCNN4	N
Potassium channel, subfamily K, member 1	KCNK1	N
Potassium channel, subfamily K, member 2	KCNK2	N
Potassium channel, subfamily K, member 3	KCNK3	N
Potassium inwardly-rectifying channel J1	KCNJ1	N
Potassium inwardly-rectifying channel J11	KCNJ11	N
Potassium voltage-gated channel A1	KCNA1	N
Potassium voltage-gated channel E1	KCNE1	N
Potassium voltage-gated channel Q1	KCNQ1	N
Potassium voltage-gated channel Q2	KCNQ2	N
Potassium voltage-gated channel Q3	KCNQ3	N
POU domain, class 1, transcription factor 1 (Pit1)	POU1F1	G
POU domain, class 3, transcription factor 4	POU3F4	G
POU domain, class 4, transcription factor 3	POU4F3	G
Pre-B-cell leukemia transcription factor 1	PBX1	G
Preproglucagon	GCG;GLP1; GLP2	G
Progesterone receptor (RU486 binding)	PGR	G

receptor)		
Prolactin	PRL	G
Prolactin receptor	PRLR	G
Proopiomelanocortin	POMC	N
Prostacyclin synthase		I
Prostaglandin 15-OH dehydrogenase	HGPD; PGDH	I
Prostaglandin D - DP receptor		I
Prostaglandin E1 receptor		I
Prostaglandin E2 receptor		I
Prostaglandin E3 receptor		I
Prostaglandin F - FP receptor		I
Prostaglandin F2 alpha receptor		I
Prostaglandin IP receptor		I
Prostaglandin-endoperoxidase synthase 2	PTGS2	G
Protease nexin 2	PN2	E
Protein C	PROC	I
Protein kinase DNA-activated	PRKDC	E
Protein S	PROS1	I
Pterin-4-alpha-carbinolamine	PCBD	
Purine nucleoside phosphorylase	NP	E
Purinergic receptor P1A1		N
Purinergic receptor P1A2		N
Purinergic receptor P1A3		N
Purinergic receptor P2X, 1	P2RX1	N
Purinergic receptor P2X, 2	P2RX2	N
Purinergic receptor P2X, 3	P2RX3	N
Purinergic receptor P2X, 4	P2RX4	N
Purinergic receptor P2X, 5	P2RX5	N
Purinergic receptor P2X, 6	P2RX6	N
Purinergic receptor P2X, 7	P2RX7	N
Purinergic receptor P2Y, 1	P2RY1	N
Purinergic receptor P2Y, 11	P2RY11	N
Purinergic receptor P2Y, 2	P2RY2	N
RAD51, DNA repair protein	RAD51	G
RAD52, DNA repair protein	RAD52	G
RAD54, DNA repair protein	RAD54	G
RAD55, DNA repair protein	RAD55	G
RAD57, DNA repair protein	RAD57	G
Recombination activating gene 1	RAG1	G
Recombination activating gene 2	RAG2	G
Red cone pigment	RCP	S
Replication factor A		E
Replication factor C	RFC2	E
Retinaldehyde binding protein 1	RLBP1	T
Retinoic acid receptor, alpha	RARA	G
Retinoic acid receptor, beta	RARB	G
Retinoic acid receptor, gamma	RARG	G
Retinoid X receptor, alpha	RXRA	G

Retinoid X receptor, beta	RXRB	G
Retinoid X receptor, gamma	RXRG	G
Retinol binding protein 1		T
Retinol binding protein 2		T
Retinol binding protein 4	RBP4	T
Ribonucleotide reductase, RRM		E
Ribosephosphate pyrophosphokinase		E
Ribosomal protein L13A	RPL13A	G
Ribosomal protein S19	RPS19	E
Ribosomal protein S4, X-linked	RPS4X	E
Ribosomal protein S6 kinase	RPS6KA3	E
Ribosomal protein S9	RPS9	G
S-adenosylmethionine decarboxylase, AMD		E
Secretin	SCT	T
Secretin receptor, SCTR	SCTR	T
Serine hydroxymethyltransferase	SHMT	E
Serotonin N-acetyltransferase	SNAT	N
Serotonin receptor, 5HT1A	HTR1A	N
Serotonin receptor, 5HT1B	HTR1B	N
Serotonin receptor, 5HT1C	HTR1C	N
Serotonin receptor, 5HT1D	HTR1D	N
Serotonin receptor, 5HT1E	HTR1E	N
Serotonin receptor, 5HT1F	HTR1F	N
Serotonin receptor, 5HT2A	HTR2A	N
Serotonin receptor, 5HT2B	HTR2B	N
Serotonin receptor, 5HT2C	HTR2C	N
Serotonin receptor, 5HT3	HTR3	N
Serotonin receptor, 5HT4	HTR4	N
Serotonin receptor, 5HT5	HTR5	N
Serotonin receptor, 5HT6	HTR6	N
Serotonin receptor, 5HT7	HTR7	N
Slug protein		G
Small nuclear ribonucleoprotein polypeptide N	SNRPN	S
Sodium channel, non-voltage gated 1, alpha	SCNN1A	N
Sodium channel, non-voltage gated 1, beta	SCNN1B	N
Sodium channel, non-voltage gated 1, gamma	SCNN1G	N
Sodium channel, voltage gated, type IV, alpha polypeptide	SCN4A	N
Sodium channel, voltage gated, type V, alpha polypeptide	SCN5A	N
Sodium channel, voltage-gated, type 1, beta polypeptide	SCN1B	N
Solute carrier family 1 (amino acid transporter), member 6	SLC1A6	T
Solute carrier family 1 (glial high affinity glutamate transporter), member 3	SLC1A3	T
Solute carrier family 1 (glutamate transporter), member 1	SLC1A1	T

Solute carrier family 1 (glutamate transporter), member 2	SLC1A2	T
Solute carrier family 1 (neutral amino acid transporter), member 4	SLC1A4	T
Solute carrier family 10 (sodium/bile acid cotransporter family), member 1	SLC10A1	T
Solute carrier family 10 (sodium/bile acid cotransporter family), member 2	SLC10A2	T
Solute carrier family 12, member 1	SLC12A1	T
Solute carrier family 12, member 2	SLC12A2	T
Solute carrier family 12, member 3	SLC12A3	T
Solute carrier family 14, member 2	SLC14A2	T
Solute carrier family 15 (H ⁺ /peptide transporter, intestinal), member 1	SLC15A1	T
Solute carrier family 15 (H ⁺ /peptide transporter, kidney), member 2	SLC15A2	T
Solute carrier family 16 (monocarboxylate transporter), member 1	SLC16A1	T
Solute carrier family 16 (monocarboxylate transporter), member 7	SLC16A7	T
Solute carrier family 17, member 1	SLC17A1	T
Solute carrier family 17, member 2	SLC17A2	T
Solute carrier family 18, member 3	SLC18A3	T
Solute carrier family 19 (folate transporter), member 1	SLC19A1	T
Solute carrier family 2 (facilitated glucose transporter), member 1	SLC2A1	T
Solute carrier family 2 (facilitated glucose transporter), member 2	SLC2A2	T
Solute carrier family 2 (facilitated glucose transporter), member 3	SLC2A3	T
Solute carrier family 2 (facilitated glucose transporter), member 4	SLC2A4	T
Solute carrier family 2 (facilitated glucose transporter), member 5	SLC2A5	T
Solute carrier family 20, member 1	SLC20A1	T
Solute carrier family 20, member 2	SLC20A2	T
Solute carrier family 20, member 3	SLC20A3	T
Solute carrier family 21, member 2	SLC21A2	T
Solute carrier family 21, member 3	SLC21A3	T
Solute carrier family 22, member 1	SLC22A1	T
Solute carrier family 22, member 2	SLC22A2	T
Solute carrier family 22, member 5	SLC22A5	T
Solute carrier family 25, member 12	SLC25A12	T
Solute carrier family 3 (facilitated glucose transporter), member 1	SLC3A1	T
Solute carrier family 4 (anion exchanger), member 1	SLC4A1	T

Solute carrier family 4 (anion exchanger), member 2	SLC4A2	T
Solute carrier family 4 (anion exchanger), member 3	SLC4A3	T
Solute carrier family 5 (sodium/glucose transporter), member 1	SLC5A1	T
Solute carrier family 5 (sodium/glucose transporter), member 2	SLC5A2	T
Solute carrier family 5 (sodium/glucose transporter), member 5	SLC5A5	T
Solute carrier family 5, member 3	SLC5A3	T
Solute carrier family 6 (GAMMA-AMINOBUTYRIC ACID transporter), member 1	SLC6A1	T
Solute carrier family 6 (neurotransmitter transporter, dopamine), member 3	SLC6A3	T
Solute carrier family 6 (neurotransmitter transporter, noradrenaline), member 2	SLC6A2	T
Solute carrier family 6 (neurotransmitter transporter, serotonin), member 4	SLC6A4	T
Solute carrier family 6, member 10	SLC6A10	T
Solute carrier family 6, member 6	SLC6A6	T
Solute carrier family 6, member 8	SLC6A8	T
Solute carrier family 7(amino acid transporter), member 1	SLC7A1	T
Solute carrier family 7(amino acid transporter), member 2	SLC7A2	T
Solute carrier family 7(amino acid transporter), member 7	SLC7A7	T
Solute carrier family 8 (sodium/calcium exchanger), member 1	SLC8A1	T
Somatostatin	SST	N
Somatostatin receptor, SSTR1	SSTR1	N
Somatostatin receptor, SSTR2	SSTR2	G
Somatostatin receptor, SSTR3	SSTR3	N
Somatostatin receptor, SSTR4	SSTR4	N
Somatostatin receptor, SSTR5	SSTR5	N
Sorcin	SRI	T
SOS1 guanine nucleotide exchange factor	SOS1	G
Steroid 5 alpha reductase 1	SRD5A1	E
Steroid 5 alpha reductase 2	SRD5A2	E
Steroid hormone receptor responsive DNA elements		G
Sterol carrier protein 2	SCP2	T
Succinic semi-aldehyde dehydrogenase	ssadh	E
Sucrase		E
Sulfonylurea receptor	SUR	G
Synaptic vesicle amine transporter	SVAT	N
Tachykinin receptor, NK1R	TACR1	N

Tachykinin receptor, NK2R	TACR2	N
Tachykinin receptor, NK3R	TACR3	N
Terminal deoxynucleotidyltransferase	TDT	I
Thiopurine S-methyltransferase	TPMT	E
Thrombopoietin	THPO	G
Thromboxane A synthase 1	TBXAS1	I
Thromboxane A2	TXA2	I
Thromboxane A2 receptor	TBXA2R	I
Thymidylate synthase	TYMS	E
Thymopoietin	TMPO	G
Thyroid hormone receptor, beta	THRB	G
Thyroid-stimulating hormone receptor	TSHR	G
Thyroid-stimulating hormone, alpha	TSHA	G
Thyroid-stimulating hormone, beta	TSHB	G
Topoisomerase I		E
Topoisomerase II		E
Transcription factor 1, hepatic	TCF1	G
Transcription factor 2, hepatic	TCF2	G
Transcription factor 3	TCF3	G
Transcription factor binding to IGHM enhancer 3	TFE3	G
Transcription factor, TUPLE1	TUPLE1	N
Transcription termination factor, RNA polymerase 1	TTF1	G
Transcription termination factor, RNA polymerase 2	TTF2	G
Transcription termination factor, RNA polymerase 3	TTF3	G
Transferrin	TF	G
Transferrin receptor	TFRC	G
Transthyretin	TTR	T
Tubulin		S
Tumour necrosis factor (TNF) receptor associated factor 1	TRAF1	I
Tumour necrosis factor (TNF) receptor associated factor 2	TRAF2	I
Tumour necrosis factor (TNF) receptor associated factor 3	TRAF3	I
Tumour necrosis factor (TNF) receptor associated factor 4	TRAF4	I
Tumour necrosis factor (TNF) receptor associated factor 5	TRAF5	I
Tumour necrosis factor (TNF) receptor associated factor 6	TRAF6	I
Tumour necrosis factor alpha	TNFA	I
Tumour necrosis factor alpha receptor	TNFAR	I
Tumour necrosis factor beta	TNFB	I
Tumour necrosis factor beta receptor	TNFBR	I

Tumour protein p53	TP53, P53	G
Tumour protein p63	TP63	G
Tumour suppressor gene DRA	DRA	I
Ubiquitin		G
Ubiquitin activating enzyme, E1		E
Ubiquitin B	UBB	G
Ubiquitin C	UBC	G
Ubiquitin carboxyl-terminal esterase L1	UCHL1	G
Ubiquitin protein ligase E3A	UBE3A	E
UDP-glucose pyrophosphorylase		E
UDP-glucuronosyltransferase 1	ugt1d, UGT1	E
UDP-glucuronosyltransferase 2	UGT2	E
Uncoupling protein 1		T
Uncoupling protein 3	UCP3	T
Uridine monophosphate kinase	UMPK	I
Uridine monophosphate synthetase	UMPS	I
Uridinediphosphate(UDP)-galactose-4-epimerase	GALE	E
Vimentin	VIM	I
Vitamin B12-binding (R) protein		G
Vitamin D receptor	VDR	G
Xanthine dehydrogenase	XDH	E
Xeroderma pigmentosum, complementation group A	XPA	E
Xeroderma pigmentosum, complementation group B	XPB	E
Xeroderma pigmentosum, complementation group C	XPC	E
Xeroderma pigmentosum, complementation group D		E
Xeroderma pigmentosum, complementation group E		E
Xeroderma pigmentosum, complementation group F	XPF	E
Xeroderma pigmentosum, complementation group G	ERCC5	E
X-ray repair gene	XRCC9	G
Xylitol dehydrogenase		E
YY1 transcription factor	YY1	G

2. A set of probes, said probes being antibodies or antibody fragments which interact with specific expressed proteins encoded by gene sequences of a group of genes, said probes being for detecting relevant variants (mutations and polymorphisms), e.g. nucleotide substitutions (missense, nonsense, splicing and regulatory), small deletions, small insertions, small insertion deletions, gross insertions, gross deletions, duplications, complex rearrangements and repeat variations in a target group of genes; characterised in that said group is a core group of genes consisting of substantially all of the genes defined in claim

- 1.
3. A set according to claim 1 or 2 in which a minority of said probes for listed genes are absent.
4. A set according to claim 1 or 2 in which a limited number of additional probes are present together with substantially all of the probes for the listed genes.
5. A set according to claim 1 or 2 in which a limited number of probes are replaced by probes for non-listed genes.
6. A set of probes for a core group of genes according to any of claims 1 to 5 in which each gene to be probed is substantially similar (greater than 85% homologous) in sequence to the respective member of the core list of genes.
7. A set according to any of claims 1 to 6 consisting of probes for members of a sub-group of the core group.
8. A set according to any preceding claim in which said probes are in the form of an array and are spatially arranged at known locations on a substrate.
9. A set according to any preceding claim wherein said probes are on a substrate which forms part of or consists of one or more chip plate(s), for use in a chip assay for detection of said gene variants.
10. A set according to any preceding claim in which said probes are mass, electrostatic or fluorescence tagged probes.
11. A set according to claim 8 or 9 in which said substrate is a semiconductor microchip.
12. A set according to any preceding claim for use in a biological assay for detection of said gene variants.
13. A set according to any preceding claim for use in the measurement of differential gene expression levels.
14. A medical device including a set according to any preceding claim for use in an assay for detection of said gene variants.
15. A medical device including a set according to any of claims 1 to 13 for use in an array for detection of differential gene expression levels.
16. A method for use in assessing the genomic profile of a patient or individual, the method comprising testing for and detecting the presence or absence of DNA or RNA encoding the relevant structural variants (as defined in claim 1) in a target group of genes by hybridising a nucleic acid-containing sample from said patient or individual to a set according to any of claims 1 and 3 to 13 and relating the probe hybridisation pattern to said variations.
17. A method for use in assessing the the genomic profile of a patient or individual, the method comprising testing for and detecting the presence or absence of DNA or RNA encoding the relevant structural variants (as defined in claim 2) in a target group of genes by interacting an expressed-protein-containing sample from said patient or individual with a set of probes according to any of claims 2 to 13 and relating the probe interaction pattern to said variations.
18. Use of a set or device according to any of claims 1 to 13 for the prognosis and management of patients suffering from or at risk of adverse events.

19. Use of a set or device according to any of claims 1 to 13 for predicting likely therapeutic response and adverse events following therapeutic intervention.
20. Use of a set or device according to any of claims 1 to 13 for predicting likely therapeutic response and adverse events following the intake of a specific drug.
20. Use of a set or device according to any of claims 1 to 13 for predicting likely patterns of symptom clusters (symptom profiles) in disease and the likelihood of subsequent, contingent, disease or symptoms.
21. Use of a set or device according to any of claims 1 to 13 for general health screening, occupational health purposes, healthcare planning on a population basis and other healthcare management utilisations.
22. Use of a set or device according to any of claims 1 to 13 for the development of new strategies of therapeutic intervention and in clinical trials.
23. Use of a set or device according to any of claims 1 to 13 for construction of and generation of algorithms for patient and healthcare management.
24. Use of a set or device according to any of claims 1 to 13 for modelling or assessing the impact of diseases or healthcare management strategies on individuals, groups, patient cohorts or populations
25. Use of a set or device according to any of claims 1 to 13 for modelling, assessing or exploring the theoretical impact of diseases and healthcare management strategies on individuals, groups, patient cohorts or populations.
26. Use of a set or device according to any of claims 1 to 13 for predicting optimum configuration/management of thereapeutic intervention.
27. A method according to claim 16 or 17 in which the identification of gene variants is indicative of a higher risk of experiencing adverse events for the patient or individual.
28. A method for generating a model to assess whether a patient or individual or population or group is or are likely to experience adverse events, which method comprises:
 - i) obtaining DNA or RNA or protein samples from patients or individuals diagnosed as suffering from adverse events;
 - ii) obtaining DNA or RNA or protein samples from a control group of subjects diagnosed as not suffering from the adverse events;
 - iii) analysing the samples obtained in i) and ii) to identify the polymorphic variations encoded in the core group of genes as defined in any of claims 1 to 7;
 - iv) calculating the frequencies of these alleles in the samples from i) and ii);
 - v) comparing the frequencies of these alleles in i) and ii);
 - vi) performing a statistical analysis on the results from v) in order to generate a model for assessing the risk of experiencing adverse events.
29. A method for assessing whether a given subject will be at risk of developing symptoms, which comprises comparing said subject's genotype with a model generated by the method of claim 28.
30. A method according to any of claims 16, 17, 28 and 29 wherein at least one step is computer-controlled.
31. An assay suitable for use in a method according to any of claims 16, 17, 28 and 29; said assay comprising means for determining the presence or absence of relevant polymorphic variants of the core group of genes as defined in any of claims 1 to 7 in a biological sample.

32. A formatted assay technique (kit) for use in assessing the risk of a patient or individual experiencing adverse events; said kit comprising:
- i) means for testing for the presence or absence of DNA or RNA encoding relevant polymorphic variants of the core group of genes as defined in claim 1 or 3 to 7 in a sample of human DNA;
 - ii) reagents for use in the detection process
 - iii) readout indicating the probability of a patient or individual experiencing adverse events.
33. A formatted assay technique (kit) for use in assessing the risk of a patient or individual experiencing adverse events; said kit comprising:
- i) means for testing for the presence or absence of proteins encoded by the core group of genes and/or relevant polymorphic variants of the core group of genes as defined in any of claims 2 to 7 in an expressed-protein-containing human sample;
 - ii) reagents for use in the detection process
 - iii) readout indicating the probability of a patient or individual experiencing adverse events.
34. A set of probes according to claim 1, wherein the probes are selected from the group consisting of oligonucleotides and polynucleotides.
35. A set of nucleotide probes for detecting relevant variants (mutations and polymorphisms), e.g. nucleotide substitutions (missense, nonsense, splicing and regulatory), small deletions, small insertions, small insertion deletions, gross insertions, gross deletions, duplications, complex rearrangements and repeat variations in a target group of genes which relate to cancer; said probes being complementary to DNA and RNA sequences of said group of genes; characterised in that said group is a core group of genes consisting of substantially all of the following:

KEY TO 'PROTEIN FUNCTION' COLUMN

E ENZYME
 T TRANSPORT & STORAGE
 S STRUCTURAL
 I IMMUNITY
 N NERVOUS TRANSMISSION
 G GROWTH & DIFFERENTIATION

ONCOLOGY GENE LIST

	HUGO gene symbol	Protein function
Absent in melanoma 1 gene	AIM1	G
Actin, alpha, cardiac	ACTC	S
Actin, alpha, skeletal	ACTA1	S
Actin, alpha, smooth, aortic	ACTA2	S
Activin		G
Activin A receptor, type 2B	ACVR2B	G
Activin A receptor, type 2-like kinase 1	ACVRL1	G
Adenomatous polyposis coli tumour suppressor	APC	G

Wilms tumour gene 4	WT4	G
Winged helix nude	WHN	G
Wiskott-Aldrich syndrome protein	WASP, THC	I
Xeroderma pigmentosum, complementation group B	XPB	E
Xeroderma pigmentosum, complementation group C	XPC	E
Xeroderma pigmentosum, complementation group D		E
Xeroderma pigmentosum, complementation group E		E
Xeroderma pigmentosum, complementation group F	XPF	E
Xeroderma pigmentosum, complementation group G	ERCC5	E
X-ray repair gene	XRCC9	G
YY1 transcription factor	YY1	G
Zinc finger protein 198	ZIC198	S
Zinc finger protein HRX	ALL1	I

36. A set of probes, said probes being antibodies or antibody fragments which interact with specific expressed proteins encoded by gene sequences of a group of genes, said probes being for detecting relevant variants (mutations and polymorphisms), e.g. nucleotide substitutions (missense, nonsense, splicing and regulatory), small deletions, small insertions, small insertion deletions, gross insertions, gross deletions, duplications, complex rearrangements and repeat variations in a target group of genes; characterised in that said group is a core group of genes consisting of substantially all of the genes defined in claim 35.
37. A set according to claim 35 or 36 in which a minority of said probes for listed genes are absent.
38. A set according to claim 35 or 36 in which a limited number of additional probes are present together with substantially all of the probes for the listed genes.
39. A set according to claim 35 or 36 in which a limited number of probes are replaced by probes for non-listed genes.
40. A set of probes for a core group of genes according to any of claims 35 to 39 in which each gene to be probed is substantially similar (greater than 85% homologous) in sequence to the respective member of the core list of genes.
41. A set according to any of claims 35 to 40 consisting of probes for members of a sub-group of the core group.
42. A set according to any preceding claim in which said probes are in the form of an array and are spatially arranged at known locations on a substrate.

43. A set according to any preceding claim wherein said probes are on a substrate which forms part of or consists of one or more chip plate(s), for use in a chip assay for detection of said gene variants.
44. A set according to any preceding claim in which said probes are mass, electrostatic or fluorescence tagged probes.
45. A set according to claim 42 or 43 in which said substrate is a semiconductor microchip.
46. A set according to any preceding claim for use in a biological assay for detection of said gene variants.
47. A set according to any preceding claim for use in the measurement of differential gene expression levels.
48. A medical device including a set according to any preceding claim for use in an assay for detection of said gene variants.
49. A medical device including a set according to any of claims 35 to 47 for use in an array for detection of differential gene expression levels.
50. A method for use in assessing the genomic profile of a patient or individual, the method comprising testing for and detecting the presence or absence of DNA or RNA encoding the relevant structural variants (as defined in claim 35) in a target group of genes by hybridising a nucleic acid-containing sample from said patient or individual to a set according to any of claims 35 and 37 to 47 and relating the probe hybridisation pattern to said variations.
51. A method for use in assessing the the genomic profile of a patient or individual, the method comprising testing for and detecting the presence or absence of DNA or RNA encoding the relevant structural variants (as defined in claim 36) in a target group of genes by interacting an expressed-protein-containing sample from said patient or individual with a set of probes according to any of claims 36 to 47 and relating the probe interaction pattern to said variations.
52. Use of a set or device according to any of claims 35 to 47 for the prognosis and management of patients suffering from or at risk of developing symptoms and consequences of cancer.
53. Use of a set or device according to any of claims 35 to 47 for predicting likely therapeutic response and adverse events following therapeutic intervention.
54. Use of a set or device according to any of claims 35 to 47 for predicting likely therapeutic response and adverse events following the intake of a specific drug.
55. Use of a set or device according to any of claims 35 to 47 for predicting likely patterns of symptom clusters (symptom profiles) in disease and the likelihood of subsequent, contingent, disease or symptoms.
56. Use of a set or device according to any of claims 35 to 47 for general health screening, occupational health purposes, healthcare planning on a population basis and other healthcare management utilisations.
57. Use of a set or device according to any of claims 35 to 47 for the development of new strategies of therapeutic intervention and in clinical trials.
58. Use of a set or device according to any of claims 35 to 47 for construction of and generation of algorithms for patient and healthcare management.
59. Use of a set or device according to any of claims 35 to 47 for modelling or assessing the impact of diseases or healthcare management strategies on

- individuals, groups, patient cohorts or populations
60. Use of a set or device according to any of claims 35 to 47 for modelling, assessing or exploring the theoretical impact of diseases and healthcare management strategies on individuals, groups, patient cohorts or populations.
 61. Use of a set or device according to any of claims 35 to 47 for predicting optimum configuration/management of thereapeutic intervention.
 62. A method according to claim 50 or 51 in which the identification of gene variants is indicative of a higher risk of developing symptoms and consequences of cancer for the patient or individual.
 63. A method for generating a model to assess whether a patient or individual or population or group is or are likely to develop symptoms and consequences of cancer which method comprises:
 - i) obtaining DNA or RNA or protein samples from patients or individuals diagnosed as suffering from cancer;
 - ii) obtaining DNA or RNA or protein samples from a control group of subjects diagnosed as not suffering from the cancer;
 - iii) analysing the samples obtained in i) and ii) to identify the polymorphic variations encoded in the core group of genes as defined in any of claims 35 to 41;
 - iv) calculating the frequencies of these alleles in the samples from i) and ii);
 - v) comparing the frequencies of these alleles in i) and ii);
 - vi) performing a statistical analysis on the results from v) in order to generate a model for assessing the risk of developing symptoms and consequences of cancer.
 64. A method for assessing whether a given subject will be at risk of developing symptoms and consequences of cancer, which comprises comparing said subject's genotype with a model generated by the method of claim 63.
 65. A method according to any of claims 50, 51, 63 and 64 wherein at least one step is computer-controlled.
 66. An assay suitable for use in a method according to any of claims 50, 51, 63 and 64; said assay comprising means for determining the presence or absence of relevant polymorphic variants of the core group of genes as defined in any of claims 35 to 41 in a biological sample.
 67. A formatted assay technique (kit) for use in assessing the risk of a patient or individual developing symptoms and consequences of cancer; said kit comprising:
 - i) means for testing for the presence or absence of DNA or RNA encoding relevant polymorphic variants of the core group of genes as defined in claim 35 or 37 to 41 in a sample of human DNA;
 - ii) reagents for use in the detection process
 - iii) readout indicating the probability of a patient or individual developing symptoms and consequences of cancer.
 68. A formatted assay technique (kit) for use in assessing the risk of a patient or individual developing symptoms and consequences of cancer; said kit comprising:
 - i) means for testing for the presence or absence of proteins encoded by the core group of genes and/or relevant polymorphic variants of the core

- group of genes as defined in any of claims 36 to 41 in an expressed-protein-containing human sample;
- ii) reagents for use in the detection process
 - iii) readout indicating the probability of a patient or individual developing symptoms and consequences of cancer.
69. A set of probes according to claim 35, wherein the probes are selected from the group consisting of oligonucleotides and polynucleotides.
70. A set of nucleotide probes for detecting relevant variants (mutations and polymorphisms), e.g. nucleotide substitutions (missense, nonsense, splicing and regulatory), small deletions, small insertions, small insertion deletions, gross insertions, gross deletions, duplications, complex rearrangements and repeat variations in a target group of genes which relate to CNS dysfunction, damage or disease; said probes being complementary to DNA and RNA sequences of said group of genes; characterised in that said group is a core group of genes consisting of substantially all of the following:

KEY TO 'PROTEIN FUNCTION' COLUMN

E ENZYME
 T TRANSPORT & STORAGE
 S STRUCTURAL
 I IMMUNITY
 N NERVOUS TRANSMISSION
 G GROWTH & DIFFERENTIATION

CNS GENE LIST

	HUGO gene symbol	Protein function
11beta hydroxysteroid dehydrogenase 2	HSD11B2	E
2,3-bisphosphoglycerate mutase	BPGM	E
2,4-dienoyl CoA reductase	DECR	E
3 beta hydroxysteroid dehydrogenase 2	HSD3B2	E
3-oxoacid CoA transferase	OXCT	E
4-hydroxyphenylpyruvate dioxygenase	HPD	E
5,10-methylenetetrahydrofolate reductase (NADPH)	MTHFR	E
6-pyruvoyltetrahydropterin synthase	PTS	E
Acetoacetyl 2-CoA-thiolase	ACAT2	E
Acetyl CoA acyltransferase	ACAA	E
Acetyl CoA carboxylase alpha	ACACA	E
Acetylcholine receptor, nicotinic, alpha A1	CHRNA1	N
Acetylcholine receptor, nicotinic, alpha A2	CHRNA2	N
Acetylcholine receptor, nicotinic, alpha A3	CHRNA3	N
Acetylcholine receptor, nicotinic, alpha A4	CHRNA4	N
Acetylcholine receptor, nicotinic, alpha A5	CHRNA5	N
Acetylcholine receptor, nicotinic, alpha A6	CHRNA6	N
Acetylcholine receptor, nicotinic, alpha A7	CHRNA7	N
Acetylcholine receptor, nicotinic, beta 1	CHRNA1	N
Acetylcholine receptor, nicotinic, beta 2	CHRNA2	N